# CENTRAL UNIVERSITY OF PUNJAB

**Master of Science in Human Genetics**

## Batch - 2022

# Department of Human Genetics and Molecular Medicine

**Syllabus**
**M.Sc. in Human Genetics (Batch-2022)**

## Graduate attributes

The graduates of the Master of Science in Human Genetics will acquire the following:

### Context of Society

The students of this course will understand the significant role of human genetics/genomics, human cytogenetics, pharmacogenomics, nutrigenomics, therapies for genetic diseases and cell and molecular biology play in the broader societal context. There will be an understanding of the basics of professional ethics, research ethics, biosafety issues, and the principles of professional practice.

### Enterprising and Knowledgeable

The course content will develop skills in human genetics, molecular biology, and bioinformatics through the subject content across a broad range of modules among the students. The development of skills in human genetics will enhance employability in the field of human genetics on account of their inclusion in clinical practice. The emphasis is on student-centric learning where they solve the patterns of inheritance by drawing pedigrees and discuss the current therapeutic interventions to treat specific genetic disorders.

### Digital and research methodology-based skills

The students will be able to study and learn the effective use of digital tools to support academic writing, reference management and independent study using digital resources and learning materials. The understanding of the principles of experimental design and methods will help the students to explore human genetics relevant research areas.

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# Course Structure of the Programme

**Total Credit: 86**

| | Core Subjects | Elective Courses | | | Foundation Courses | | Total Credit |
|---|---|---|---|---|---|---|---|
| | | DE | ID | SB | CF | EF/VB | |
| Sem-I | 03 (9 Cr) | 01 (3 Cr) | 01 (2 Cr) | 03 (3 Cr) | 01 (3 Cr) | -- | **20** |
| Sem-II | 04 (12 Cr) | 01 (3 Cr) | -- | 02 (4 Cr) | 01 (3 Cr) | 01 (2 Cr) | **24** |
| SEM-III | 03 (9 Cr) | 01 (3 Cr) | -- | 01 (3 Cr)<br>01 (4 Cr Dissertation) | 01 (2 Cr)<br>01 (1 Cr) | -- | **22** |
| SEM-IV | -- | -- | -- | 01 (20 Cr Dissertation) | -- | -- | **20** |
| **Credit Score** | **30** | **9** | **02** | **34** | **9** | **02** | **86** |

**DE:** Discipline Elective
**ID:** Interdisciplinary
**SB:** Skill based (Practicals); Dissertation
**CF:** Compulsory foundation
**EF:** Elective Foundation
**VB**: Value Based

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# Semester-I

| Course Code | Course Title | Course Type | Hours | | | Credit |
|---|---|---|---|---|---|---|
| | | | L | T | P | |
| MME.506 | Cell Biology | Core | 3 | 0 | 0 | 3 |
| HGE.507 | Concepts of Genetics | Core | 3 | 0 | 0 | 3 |
| HGE.508 | Basic and Clinical Biochemistry | Core | 3 | 0 | 0 | 3 |
| HGE.509 | Biostatistics and Research Methodology | Compulsory Foundation | 3 | 0 | 0 | 3 |
| HGE.510 | Concepts of Genetics (Practical) | Skill based | 0 | 0 | 2 | 1 |
| HGE.511 | Basic and Clinical Biochemistry (Practical) | Skill based | 0 | 0 | 2 | 1 |
| HGE.512 | Biostatistics and Research Methodology (Practical) | Skill based | 0 | 0 | 2 | 1 |
| **Discipline Elective Course-I (Any one of the following)** | | | | | | |
| HGE.515 | Population Genetics and Genetic Epidemiology | DE | 3 | 0 | 0 | 3 |
| MME.515 | Molecular and Cellular Oncology | DE | 3 | 0 | 0 | 3 |
| ZOL.525 | Nanobiology | DE | 3 | 0 | 0 | 3 |
| BIM.511 | Protein Engineering | DE | 3 | 0 | 0 | 3 |
| **Interdisciplinary Course-I (For other Departments)** | | | | | | |
| HGE.518 | Introduction to Intellectual Property Rights | IDC | 2 | 0 | 0 | 2 |
| XXX. | Choose from IDC courses offered by other Departments | IDC | 2 | 0 | 0 | 2 |
| | | | | | **Total Credits** | **20** |

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## Semester-II

| Course Code | Course Title | Course Type | Hours | | | Credit |
|---|---|---|---|---|---|---|
| | | | L | T | P | |
| HGE.521 | Human Cytogenetics and Human Biochemical Genetics | Core | 3 | 0 | 0 | 3 |
| MME.522 | Essentials of Immunology | Core | 3 | 0 | 0 | 3 |
| HGE.523 | Advanced Techniques in Human Genetics | Core | 3 | 0 | 0 | 3 |
| HGE.524 | Human Anatomy and Physiology | Core | 3 | 0 | 0 | 3 |
| MME.525 | Essentials of Immunology (Practical) | Skill Based | 0 | 0 | 2 | 1 |
| HGE.526 | Clinical Genetics (Practical) | Skill Based | 0 | 0 | 6 | 3 |
| MME.528 | Molecular Biology | Compulsory Foundation | 3 | 0 | 0 | 3 |
| HGE.529 | Principles of Ecological Science | Value based | 2 | 0 | 0 | 2 |
| XXX | Value Added Course (From other departments) | VAC | 2 | 0 | 0 | 2 |
| **Discipline Elective Course-II (Any one of the following)** | | | | | | |
| HGE.527 | Human Embryology and Developmental Genetics | DE | 3 | 0 | 0 | 3 |
| MME.527 | Stem Cell and Regenerative Medicine | DE | 3 | 0 | 0 | 3 |
| MIC.525 | Microbial Pathogenicity | DE | 3 | 0 | 0 | 3 |
| ZOL.553 | Vascular Biology | DE | 3 | 0 | 0 | 3 |
| BIM.521 | Big Data Analysis in Bioinformatics and Healthcare | DE | 3 | 0 | 0 | 3 |
| ZOL.554 | Neurobiology and Degenerative Pathophysiology | DE | 3 | 0 | 0 | 3 |
| | | | | | **Total credits** | **24** |

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# Semester-III

| Course Code | Course Title | Course Type | Hours | | | Credit |
|---|---|---|---|---|---|---|
| | | | L | T | P | |
| HGE.551 | Pharmacogenomics and Nutrigenomics | Core | 3 | 0 | 0 | 3 |
| HGE.552 | Genetic Diseases and Therapies | Core | 3 | 0 | 0 | 3 |
| HGE.553 | Recombinant DNA technology and therapeutics | Core | 3 | 0 | 0 | 3 |
| MME.554 | Tools in Bioinformatics (Practical) | Skill Based | 0 | 0 | 6 | 3 |
| **Discipline Elective Course-III (Any one of the following)** | | | | | | |
| HGE.555 | Biosafety, Bioethics and Intellectual Property Rights | DE | 3 | 0 | 0 | 3 |
| MME.555 | Evolution and Developmental Biology | DE | 3 | 0 | 0 | 3 |
| **Foundation Courses** | | | | | | |
| | | | | | | |
| MME.557 | Concepts of Bioinformatics | Discipline Enrichment | 2 | 0 | 0 | 2 |
| HGE.558 | Innovation and Entrepreneurship | Compulsory Foundation | 1 | 0 | 0 | 1 |
| HGE.600 | Dissertation Part-I | Skill Based | 0 | 0 | 8 | 4 |
| **Total Credits** | | | | | | **22** |

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## Semester-IV

| Course Code | Course Title | Course Type | Hours | | | Credit |
|---|---|---|---|---|---|---|
| | | | L | T | P | |
| HGE.601 | Dissertation Part-II | Skill Based | 0 | 0 | 40 | 20 |
| Total | | | 0 | 0 | 0 | 20 |

**MOOC Options:** 40% credits can be obtained through MOOCs depending on availability of course with matching content.

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# Semester – I

**Course Code:** **MME.506**
**Course Title:** **Cell Biology**
**Total Hours:** **45**

| L | T | P | C |
|---|---|---|---|
| 3 | 0 | 0 | 3 |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:
CLO1: Understanding structures and functions of various cellular organelles.
CLO2: Conceptualization of basic cellular mechanisms.
CLO3: Conceptualize the mechanisms of inter- as well as intra-cellular communications.
CLO4: Understanding the cell cycle regulation and its importance in disease biology

| | |
|---|---|
| **Unit 1** **12 Hours**<br>Introduction to the cell: Models of membrane structure, Membrane proteins, Membrane carbohydrates, Membrane transport of small molecules, Membrane transport of macromolecules and particles. Structural organization and function of intracellular organelles: The lysosomes, Ribosomes, The peroxisomes, The Golgi apparatus, The endoplasmic reticulum, Mitochondria. | **CLO1** |
| **Unit 2** **10 Hours**<br>Protein secretion and sorting: Protein secretion, synthesis and targeting to mitochondria, chloroplast, peroxisomal proteins, translational modification in the ER. Intracellular traffic, vesicular traffic in the secretary pathway, protein sorting in the Golgi bodies, traffic in the endocytic pathway, exocytosis | **CLO2** |
| **Unit 3** **14 Hours**<br>The cytoskeleton: The nature of cytoskeleton, Intermediate filaments, Microtubules, Actin filaments, Cilia and centrioles, Organization of the cytoskeleton. Cell communication and cell signaling: Cell adhesions, Cell junctions and the extra cellular matrix, Cell-cell adhesion and communication, Cell matrix adhesion, Collagen the fibrous protein of the matrix, Non-collagen component of the extra cellular matrix | **CLO3** |
| **Unit 4** **9 Hours**<br>Cell growth and division: Overview of the cell cycle and its control, molecular mechanisms for regulating mitotic and meiotic events, Amitosis, Cell cycle control, Checkpoints in cell cycle regulation. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Lecture cum demonstration; Problem solving; Self-learning.
**Internal assessment shall be through any of the following:** Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, presentations, and discussions.

**Suggested Reading:**

1. Alberts, B., Bray, D., Lews, J., Raff, M., Roberts, K. and Watson, J.D. (2010). Molecular Biology of the cell. Garland publishers, Oxford.
2. Celis, J.E. (2006). Cell biology: A laboratory handbook, Vol 1, 2, 3. Academic Press, UK.
3. Gupta, P.K. (2008). Cytology, Genetics and Evolution. Rastogi publications, Meerut, India.
4. Karp, G. (2010). Cell and Molecular Biology: Concepts and Experiments. John Wiley & Sons. Inc. New Delhi, India.

**Course Code:** **HGE.507**
**Course Title:** **Concepts of Genetics**
**Total Hours:** **45**

| L | T | P | C |
|---|---|---|---|
| 3 | 0 | 0 | 3 |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:
CLO1: Evaluate the Mendelian and Non-Mendelian inheritance patterns. Gather knowledge about gene expression regulation and sex determination,
CLO2: Evaluate different chromosomal aberrations and ploidies
CLO3: Know about genetic system of microbes
CLO4: Know the details of extra chromosomal inheritance patterns

| | |
|---|---|
| **UNIT I**<br>**11 Hours**<br>**Transmission Genetics**: Mendel's laws of inheritance and its applications; concept of segregation, independent assortment and dominance; pedigree analysis; epistasis; crossing over and recombination; gene linkage and genetic mapping<br>**Sex determination**: Sex determination in Human and Drosophila; X-chromosome inactivation; dosage compensation. | **CLO1** |
| **UNIT II** **12 Hours**<br>**Chromosomal Mutations**: Chromosomal aberrations: structural and numerical; evolutionary history of bread wheat; aneuploids–nullisomics, monosomics, and trisomics; somatic aneuploids; changes in chromosome structure; properties of chromosomes for detection of structural changes; mutations and it's types; complementation and recombination; transposable elements in Pro- and Eukaryotes.<br>**Genes and genome dynamics**: Fine structure of gene; and analysis, Benzer's experiments. | **CLO2** |
| **UNIT III** **11 Hours**<br>**Microbial Genetics:** Genetic systems of Viruses and Bacteria; genetic analysis and mapping in Bacteria and Bacteriophages – conjugation, transformation and transduction; recombination and gene mapping; evolution of microbial genome. | **CLO3** |
| **UNIT IV** **11** Hours<br>**Extra-chromosomal inheritance:** Chloroplast: variegation in Four O' Clock plants; mutations in *Chlamydomonas*; mitochondrial inheritance: poky in neurospora, petites in yeast; molecular organization and gene products of chloroplast and mitochondrial DNA; infectious heredity: Kappa in *Paramecium*: Infective particles in *Drosophila*; endosymbiont theory. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Lecture cum demonstration; Problem solving; Self-learning.

**Internal assessment shall be through any of the following:** Report on novel chromosome aberrations in human, Discussion on transposable elements in human diseases, Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, Karyotype analysis presentations and discussions.

**Suggested Readings:**

1. Klug WS and Cummings MR. Concepts of Genetics. Prentice-Hall.2014
2. Anthony, J.F. Miller, J.A. Suzuki, D.T., Richard, R.C., Gilbert, W.M. (1998). An introduction to Genetic Analysis. W.H. Freeman publication, USA.
3. Pierce BA. Genetics: A Conceptual approach. Freeman Publishers.
4. Hartle DL and Jones EW. Genetics: Analysis of Genes and Genomes. Jones & Bartett.
5. Atherly, A.G., Girton, J.R., Mcdonald, J.F. (1999). The Science of Genetics. Saundern College publication.
6. Snusted, D.P., Simmons, M. J. (2010). Principles of Genetics. John Wiley & Sons, New
7. Griffith AF et al. An Introduction to Genetic Analysis. John Wiley & Sons.

| | | L | T | P | C |
|---|---|---|---|---|---|
| **Course Code:** | **HGE.508** | | | | |
| **Course Title:** | **Basic and Clinical Biochemistry** | **3** | **0** | **0** | **3** |
| **Total Hours:** | **45** | | | | |

**Course Learning Outcomes:**

On successful completion of the course the student will be able to:

CLO1: Study the importance of maintenance of pH, role of vitamins and minerals in metabolic processes.

CLO2: Compare the basic structural features of biomolecules namely carbohydrates, lipids, proteins, and vitamins.

CLO3: Elaborate the structure, and functions of proteins, and metabolism of nucleic acids.

CLO4: Conceptualize the basic features of enzyme, its regulation and kinetics of enzyme catalysed reactions.

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| **UNIT I** **11 Hours**<br>**Clinical Biochemistry:** Properties of water, Ionization of water, weak acids and weak bases, pH and buffers. Water and sodium balance, Interpretation of biochemical tests, Composition of blood, urine and cerebrospinal fluids, Vitamins and trace elements disorders. Liver and kidney function tests, Jaundice, diabetes mellitus, hypoglycemia, hypertension. | **CLO1** |
| **UNIT II** **11 Hours**<br>**Carbohydrate Metabolism**: Classification and functions, Epimers; Anomers; Mutarotation. Reactions of carbohydrates; General scheme of metabolism. Basic concepts, Glycolysis, Krebs cycle, Pentose phosphate pathway, Gluconeogenesis, Regulation of carbohydrate metabolism. Inborn errors of carbohydrate metabolism | **CLO2** |

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|---|---|
| **Lipids:** Classification and functions, Beta oxidation - Pathway and regulation. Role of acyl carnitine in fatty acyl transport. Cholesterol synthesis. Ketone bodies- Formation and utilization. Clinical features and laboratory findings in disorders of triglyceride, lipoprotein and cholesterol metabolism. | |
| **UNIT III** **11 Hours**<br>**Proteins:** Structure and function of proteins, Secondary, Tertiary and Quaternary structure, super secondary structures, Ramachandran plot. Oxygen binding proteins – Hemoglobin and myoglobin.<br>**Nucleic Acids:** Structure and functions, Nucleosides and nucleotides. Metabolism of purines and pyrimidines- Salvage and de novo pathways. | **CLO3** |
| **UNIT IV** **12 Hours**<br>**Enzymes:** Definition, Classifications and nomenclature, prosthetic groups, cofactors, Mechanism of enzyme action and properties of enzymes; Enzyme activity, Factors affecting rate of enzyme catalyzed reactions: pH, temperature, etc. ES complex formation; Michaelis-Menten equation; Determination of Km and Vmax and its significance; Turnover number; Enzyme inhibition: reversible and irreversible inhibition. Isoenzymes, catalytic antibodies, multienzyme complexes and ribozymes. | **CLO4** |

**Transactional Modes:** Lecture**;** Tutorial**;** Virtual classrooms; Lecture cum demonstration; Problem solving; Self-learning; Google Class; Microsoft teams; YouTube demonstrations etc.

**Internal assessment shall be through any of the following:** Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, presentations, and discussions.

**Suggested Readings:**

1. Berg, J.M., Tymoczko, J.L., Gatto Jr, J.R., Stryer, L. (2015) Biochemistry Eighth Edition, 2015, W.H. Freeman & Company. USA.
2. Brown, T.A. (2016). Gene Cloning and DNA Analysis: An Introduction, 7th Edition January 2016, Blackwell Publishing Professional. USA.
3. Nelson, D.L., Cox, M.M. (2017). Lehninger Principles of Biochemistry. Seventh Edition| 2017
4. Donald Voet, Charlotte W. Pratt, Judith G. Voet (2012) Principles of Biochemistry, International Student Version. John Wiley & Sons, Publisher
5. Swaminathan, R. (2011). Handbook of clinical biochemistry. 2nd edition, World Scientific Publishing Company, New Jersey, USA
6. Palmer, T., Bonner, P.L. (2007) Enzymes: biochemistry, biotechnology and clinical chemistry. Woodhead Publishing Limited.
7. Price, N.C., Stevens, L. (2003) Fundamentals of enzymology: The cell and molecular biology of catalytic proteins. Oxford University Press, USA.
8. Swaminathan, R (2011) Handbook of Clinical Biochemistry, 2nd Edition, Publisher: World Scientific
9. Murphy M.J., Srivastava R., Deans, K. (2018) Clinical Biochemistry, 6th Edition. Elsevier's USA.
10. Devlin, T.M. (2010) Textbook of Biochemistry with clinical correlation. John Wiley and Sons Publishers.

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| | |
|---|---|
| **Course Code:** | **HGE.509** |
| **Course Title:** | **Biostatistics and Research Methodology** |
| **Total Hours:** | **45** |

| L | T | P | C |
|---|---|---|---|
| 3 | 0 | 0 | 3 |

**Course Learning Outcomes:**

On successful completion of the course the student will be able to:

CLO1: Analyze and evaluate wide variety of statistical data
CLO2: Compose statistical data and summary statistics in graphical and tabular forms. Perform biological sampling and statistical analysis.
CLO3: Apply suitable statistical tools to analyze data
CLO4: Write and communicate scientific reports, projects, and publications.

| | |
|---|---|
| **UNIT I**<br>Hours: **11**<br>Overview of Biostatistics: Basic concepts of statistical data and different types of tables; graphical representation of experimental data for publication; frequency distribution; measurement of central tendency and variation. | **CLO1** |
| **UNIT II** Hours: **11**<br>Experimental design and analysis: Basics of sampling in biological studies; different types of sampling techniques; various steps in sampling; concept of data distribution in sampling; graphical representation of data; level of significance; hypothesis testing. | **CLO2** |
| **UNIT III** Hours: **11**<br>Inferential Statistics: Chi-Square test: hypothesis testing, contingency, homogeneity; student's t-test: paired and unpaired, one tailed and two tailed; one-way and two-way analysis of variance (ANOVA); correlation and regression. | **CLO3** |
| **UNIT IV** Hours: **12**<br>Study design & Technical writing: Best practices in research and technicality of research design; interpretation and report writing, e-Library; web-based literature search engines; evaluation-based development of scientific writing skill: synopsis, research paper, poster preparation and paper presentation and dissertation. | **CLO4** |

**Transactional Modes:** Lecture**;** Tutorial; Virtual classroom, Problem solving; Self-learning.
**Internal assessment shall be through any of the following:** Surprise Tests, student generated questions, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper.

**Suggested Readings:**


1. Rao, S.P.S.S. Richard, J. (2016). Introduction to biostatistics and research methods. 5th Ed. Phi Learning Pvt. Ltd. New Delhi.
2. Hoffman, J. (2015). Biostatistics for medical and biomedical practioners. 1st Ed. Academic Press, London.
3. Banerjee P.K (2014). Introduction to Biostatistics. S.Chand, New Delhi.

4. Antonisamy, B. Christopher, S. Samuel, P.P. (2011). Biostatistics: Principles and Practice. Tata McGraw Hill. New Delhi.
5. Daniel W.W (2011). Biostatistics: Basic Concepts and methodology for the health sciences. 9th Ed. John Wiley and Sons Inc, New Delhi.
6. Norman, G. and Streiner, D. (2008). Biostatistics: The Bare Essentials. (with SPSS), 3rd Edition, Decker Inc. USA.
7. Sokal, R.R. and Rohlf, F.J. (1994). Biometry: The Principles and Practices of Statistics in Biological Research. W.H. Freeman publishers, USA.

**Course Code:** **HGE.510**
**Course Title:** **Concepts of Genetics (Practical)**
**Total Hours:** **30**

| L | T | P | C |
|---|---|---|---|
| 0 | 0 | 2 | 1 |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:
1. Perform any experiments on Mendelian genetics and pedigree analysis
2. Perform linkage based genetic analysis
3. Design genetic experiments using common model organisms
4. Isolate genomic DNA for genetic analyses

**List of Practical work:**

| | | |
|---|---|---|
| 1 | Problems on Monohybrid and dihybrid ratios, Multiple alleles, Epistasis | **CLO1** |
| 2 | Inheritance patterns in Human– Numerical on Pedigree analysis- Autosomal patterns, X–linked patterns, Y–linked patterns | |
| 3 | Segregation analysis in Drosophila (Monohybrid, Dihybrid) | |
| 4 | Analysis on Linkage | **CLO2** |
| 5 | Linkage mapping | |
| 6 | Identification of inactivated X chromosome as Barr body | **CLO3** |
| 7 | Studies of a Model organism: *E. coli, C.elegans, D. melanogaster* and *D. rerio* | |
| 8 | Isolation of genomic DNA and quality control | **CLO4** |

**Transactional Modes:** Hands-on practicals; Demonstration**;** Virtual classrooms; Tutorial**;** Self-directed learning.

**Evaluation criteria for practical courses:**

- Continuous Assessment = **60 Marks**

Based on performance of the students and completion of the record book (60 divided by the number of practicals)

- Final Examination = **40 Marks**
  Subjective question = **10 Marks**
  Performing experiment = 20 Marks
  Viva voce = 10 Marks

**Course Code:** **HGE.511**

| L | T | P | C |
|---|---|---|---|

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| | | | | | |
|---|---|---|---|---|---|
| **Course Title:** | **Basic and Clinical Biochemistry (Practical)** | **0** | **0** | **4** | **2** |
| **Total Hours:** | **60** | | | | |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:

1. Prepare buffers and solutions with varied concentration.
2. Quantitative estimation of biomolecules and their role in health and disease
3. Understand the impact of various factors affecting enzyme activity
4. Importance of Clinical tests in health and disease.

**List of Practicals**

| | |
|---|---|
| **1.** Preparation of Buffers | **CLO1** |
| **2.** Extraction of DNA, and purity check by electrophoresis. | **CLO2** |
| 3. Quantitative estimation of Glucose, Cholesterol and Proteins | **CLO2** |
| 4. Assay of enzyme activity in saliva.<br>5. Effect of temperature on enzyme activity.<br>6. Effect of pH on enzyme activity. | **CLO3** |
| 7. Liver function tests-SGOT and SGPT<br>8. Determination of urea | **CLO4** |

**Transactional Modes:** Hands-on Practicals; Demonstration**;** Virtual classrooms; Tutorial**;** Self-learning.
**Internal assessment shall be through any of the following:** Lab performance, Open book techniques, Notebook writing, presentations, and group discussions.

**Suggested Readings:**


1. Rajendran, S., Dhiman, P. (2019) Biochemistry Practical Manual. Elsevier India.
2. Plummer, D. (2004) An Introduction to Practical Biochemistry. Tata McGraw Hill Publishers Co. Ltd., India
3. Gupta, P.P., Gupta, N. (2017) Essentials of Practical Biochemistry. Jaypee Brothers Medical Publishers Pvt. Ltd. India.
4. Hofmann, A., Clokie, S. (2018). Wilson and Walker's Principles and Techniques of Biochemistry and Molecular Biology. Cambridge University Press, USA
5. Murphy M.J., Srivastava R., Deans, K. (2018) Clinical Biochemistry, 6th Edition. Elsevier's USA.
6. Swaminathan, R (2011) Handbook of Clinical Biochemistry, 2nd Edition, Publisher: World Scientific


**Evaluation criteria for practical courses:**

- Continuous Assessment = **60 Marks**

Based on performance and good lab practices of the students and completion of the record book (60 divided by the number of practicals)

- Final Examination = **40 Marks**

Subjective question = 10 Marks
Performing experiment = 20 Marks
Viva voce = 10 Marks

**Course Code:** **HGE.512**
**Course Title:** **Biostatistics and Research Methodology (Practical)**
**Total Hours:** **30**

| L | T | P | C |
|---|---|---|---|
| 0 | 0 | 2 | 1 |

**Course Learning Outcomes:**

On successful completion of the course the student will be able to:

CLO1: Identify statistical data type and plot graphs using conventional tools. Perform basic statistics to check the data quality
CLO2: Test correlation and regression using two or more variables
CLO3: Perform standard parametric and non-parametric statistics on simple data
CLO4: Write and communicate scientific literatures

**List of Practical work:**

| | | |
|---|---|---|
| 1 | Plotting different types of graphs and statistical tables using MS excel, GraphPad and/or relevant tools | **CLO1** |
| 2 | Plotting normal distribution from data given | |
| 3 | Problems on central tendency, measurement of variance (standard deviation, standard error etc.) | |
| 4 | Problems of correlation | **CLO2** |
| 5 | Problems of regression | |
| 6 | Chi-square test | **CLO3** |
| 7 | Student's t-test | |
| 8 | Analysis of variance (ANOVA) | |
| 9 | Different techniques of sampling | |
| 10 | Scientific writing skill development | **CLO4** |
| 11 | Practice writing research reports, synopsis, poster etc. | |

**Transactional Modes:** Laboratory based practicals; demonstration, Problem solving; Self-learning.

**Evaluation criteria for practical courses:**

- Continuous Assessment = **60 Marks**

Based on performance and good lab practices of the students and completion of the record book (60 divided by the number of practicals)

- Final Examination = **40 Marks**
    i. Subjective question = 10 Marks
    ii. Performing experiment = 20 Marks

iii. Viva voce = 10 Marks

# Discipline Elective-I

| | | L | T | P | C |
|---|---|---|---|---|---|
| **Course Code:** | **HGE.515** | **3** | **0** | **0** | **3** |
| **Course Title:** | **Population Genetics and Genetic Epidemiology** | | | | |
| **Total Hours:** | **45** | | | | |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:
CLO1: Conceptualize the basic concepts in population genetics and apply statistics relevant to the study of genomic diversity
CLO2: Examine the major genetic and ecological processes underlying evolution and explain the process driving speciation
CLO3: Perform different epidemiological studies to identify the cause-effect relationship in variety of human traits/diseases
CLO4: Design genetic studies and perform association and linkage analysis on any relevant data.

| | |
|---|---|
| **UNIT I** **11 Hours**<br>Population dynamics and basics of epidemiology: Dynamics and conditions of the Hardy-Weinberg law; selection coefficient and fitness; heterozygous advantages; inbreeding and its consequences; mutation pressure and estimation of rates; genetic load; dynamics of migration and genetic drifts. | **CLO1** |
| **UNIT II** **11 Hours**<br>Evolution and Speciation: Emergence of evolutionary thoughts Lamarck; Darwinism – concepts of variation, adaptation, struggle, fitness and natural selection; Mendelism; spontaneity of mutations; the evolutionary synthesis; Mechanism of evolution: Adaptive radiation; isolating mechanisms; speciation; allopatricity and sympatricity; convergent evolution; sexual selection; co-evolution; Evolution of Homo sapiens | **CLO2** |
| **UNIT III** **11 Hours**<br>Fundamentals of epidemiological studies: Different types of Experimental and observational studies; basic parameters of epidemiology: frequency, occurrence, prevalence, incidence; association; causation, variation; Association studies: candidate gene association and genome-wide association studies (GWAS); systematic review and meta-analysis. | **CLO3** |
| **UNIT IV** **11 Hours**<br>Genetic variation and complex trait inheritance: Basics of genetic variations; genetic markers – SNP, CNV, Ins/dels, VNTR, STR, microsatellite; concepts of tag markers and haplotypes, linkage disequilibrium; quantitative genetic analysis; QTL and eQTL. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Case study; Virtual classrooms; seminar presentation; Problem solving; Self-learning.
**Internal assessment shall be through any of the following:**

Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, Seminars, term papers, discussions and presentations.

**Suggested Readings:**

1. Bhasker, H.V. and Kumar S (2008). Genetics. Campus Books International, New Delhi, India.
2. Cavalli-Sforza, L.L. and Bodmer, W.F. (2013). The Genetics of Human Populations. Dover Publications.
3. Hamilton M.B. (2009). Population Genetics. Wiley-Blackwell, UK.
4. Hedrick P.W.(2011). Genetics of Populations. Jones and Bartlett Publishers, Massachusetts.
5. Jobling, M., Hollox, E., Hurles, M., Kivisild, T. and Tyler-Smith, C. (2013). Human Evolutionary Genetics. Garland Science.
6. Knight, J.C. (2009). Human Genetic Diversity –Functional consequences for Health and Disease. Oxford University Press, USA.
7. Krebs, J.E, Goldstein, E.S. and Kilpatrick, S.T. (2013) Lewin's Essential Genes. Jones and Bartlett learning, USA.
8. Nielsen, R. and Slatkin, M. (2013). An Introduction to Population Genetics: Theory and Applications. Sinauer Associates, Inc.
9. Relethford, J.H. (2012). Human Population Genetics. John Wiley & Sons.
10. Snusted, D.P., Simmons, M. J. (2010). Principles of Genetics. John Wiley & Sons, New York.
11. Palmer LJ, Burton PR & Smith GD (2011). An introduction to genetic epidemiology (Policy Press, University of Bristol)
12. Dawn TM (2011). Genetic Epidemiology (Springer)
13. Austin M (2013). Genetic Epidemiology: Methods and Applications, 1st Edition (CABI Publishing).

**Course Code:** **MME.515**
**Course Title:** **Molecular and Cellular Oncology**
**Total Hours:** **45**

| L | T | P | C |
|---|---|---|---|
| 3 | 0 | 0 | 3 |

**Course Learning Outcomes:**

On successful completion of the course the student will be able to:

CLO1: Understand fundamentals of cancer.

CLO2: Gain knowledge about genetics and signal transduction involved in tumorigenesis.

CLO3: Learn about various tools used for diagnostic purposes.

CLO4: Understand basics principles of anticancer therapeutics as well as about recent developments of the field.

| **Unit: 1** **13 Hours** | **CLO1** |
|---|---|
| Fundamentals and Genetics of Cancer: Hallmarks of cancer, cancer classification, Mutagens, carcinogens and gene mutations, Chromosomal aberrations, tumor viruses | |

| | |
|---|---|
| and discovery of oncogenes, tumor suppressors and oncogenes, familial cancer syndromes, telomere regulation in cancer. | |
| **Unit: 2** **11 Hours**<br>Signal Transduction in Cancer Progression: Deregulation of Cell cycle in cancer. Cell signaling in cancer; cancer metabolism; hypoxia and metastasis, angiogenesis, tumor microenvironment. DNA damage and repair defects and their relation to cancer, cancer stem cells. | **CLO2** |
| **Unit: 3** **11 Hours**<br>Cancer Detection: General and organ specific symptoms associated with cancer, techniques for cancer detection, biomarkers for cancer detection of various stages of cancer, In-vitro assays to detect angiogenesis, metastasis, cell proliferation, mice models to study cancer (transgenic, knock-out, knock-in, xenografts and patient derived xenografts), genomic and proteomic approaches to develop better cancer markers. | **CLO3** |
| **Unit: 4** **10 Hours**<br>Cancer Therapies and Recent Advances in Cancer Research: Traditional Chemotherapies, radiotherapy, Onco-surgery, Bone marrow transplantation, stem cell therapies, Immunotherapy, combinational therapies, natural products as therapeutics, cancer vaccines, gene therapies, targeted anticancer therapies, CAR T and other new anticancer therapies. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Lecture cum demonstration; Problem solving; Self-learning.

**Internal assessment shall be through any of the following:** Surprise Tests, case analysis, simulated problem solving, classroom assignments, term paper, presentations and discussions.

**Suggested Reading:**

1. DeVita, V. T., Rosenberg, S. A., & Lawrence, T. S. (2018). DeVita, Hellman, and Rosenberg's cancer. Lippincott Williams & Wilkins.
2. Enders, G. H. (2010). Cell cycle deregulation in cancer. Humana Press, Springer science, New York.
3. Gusev, Y. (2019). Micro RNA Profiling in Cancer. Pan Standford publishing pvt.Ltd., Singapore.
4. Hiem, S., & Mitelman, F. (2019). Cancer Cytogenetics. IIIrd edition. Willey-Blackwell publishing, New Jersey.
5. Jocelyn, E. K., Elliot, S. G., Stephen, T. K. (2018). Lewin's Gene X. Jones &Barlett.
6. Wang, E. (2018). Cancer systems biology. CRC press, Taylor & Francis group, New York.
7. Weinberg, Robert A. (2015). The Biology of Cancer. New York: Garland Science

**Related Weblink**

http://www.insidecancer.org/
http://www.who.int/cancer/en/
http://www.cancer.gov/
http://www.icmr.nic.in/ncrp/cancer_reg.htm

**Syllabus**
**M.Sc. in Human Genetics (Batch-2022)**

# Interdisciplinary Courses

| | |
|---|---|
| **Course Code:** | **HGE.518** |
| **Course Title:** | **Introduction to Intellectual Property Rights** |
| **Total Hours:** | **30** |

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|---|---|---|---|
| 2 | 0 | 0 | 2 |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:
CLO1: Understand the concept and genesis of IP
CLO2: Differentiate between plagiarism and fair use of copyright material
CLO3: Understand the Patent regime of India
CLO4: Conceptualize the other forms of IP namely Trademark, Industrial Design, Trade Secret, New varieties of plant

| | |
|---|---|
| **UNIT I** **8 Hours**<br>Brief history, current status and career opportunities in IP<br>Introduction to IP, Genesis and development of concept of IPR; WIPO administered Treaties: Paris Convention, 1883, the Berne Convention, 1886, the TRIPS Agreement, 1994; the WIPO Convention, 1967; National Innovation and Startup Policy for Students and Faculty 2019, Career Opportunities in IP. | **CLO1** |
| **UNIT II** **6 Hours**<br>Copyright and Layout Design Protection<br>Copyright and related rights; Plagiarism; Fair Use of copyright material; Layout Design Protection. | **CLO2** |
| **UNIT III** **8 Hours**<br>Patent Regime in India:<br>Patents, patentability of inventions; non-patentable subject matter, Patent registration procedure in India; Protection of Traditional Knowledge, Assignment and license of patented technology; Patent filing routes for other countries: Convention Application and Patent Cooperation Treaty (PCT) application. | **CLO3** |
| **UNIT IV** **8 Hours**<br>Other forms of IP<br>Concept, Registration and term of protection: Trademark, Industrial Design, Trade Secret, Protection of New varieties of plant, Geographical Indications | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Virtual classrooms; Lecture cum demonstration; Problem solving; Self-learning, Class activity based

**Internal assessment shall be through any of the following:** Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, term paper, Seminars.

**Suggested Readings:**
1. Dutfield G. (2003). Intellectual Property Rights and the Life Science Industries: A Twentieth Century History (Globalization and Law). Routledge.

2. Mahop, M.T. (2010). Intellectual Property, Community Rights and Human Rights: The Biological and Genetic Resources of Developing Countries. Routledge.
3. Khor M. (2002). Intellectual Property, Biodiversity and Sustainable Development: Resolving the Difficult Issues. Zed Books limited.
4. Ahuja, V.K. (2017). Law relating to Intellectual Property Rights. LexisNexis, India. 3rd Edition.
5. Mahop, M.T. (2010). Intellectual Property, Community Rights and Human Rights: The Biological and Genetic Resources of Developing Countries. Routledge, USA.
6. Neeraj, P. and Khusdeep, D. (2014). Intellectual Property Rights. India, IN: PHI learning Private Limited.
7. Nithyananda, K V. (2019). Intellectual Property Rights: Protection and Management. India, IN: Cengage Learning India Private Limited.

**Weblinks:**

1. World Intellectual Property Organization (https://www.wipo.int/about-ip/en/)
2. Office of the Controller General of Patents, Designs & Trademarks (http://www.ipindia.nic.in/)

# Semester – II

| | | L | T | P | C |
|---|---|---|---|---|---|
| **Course Code:** | **HGE.521** | | | | |
| **Course Title:** | **Human Cytogenetics and Human Biochemical Genetics** | **3** | **0** | **0** | **3** |
| **Total Hours:** | **45** | | | | |

**Course Learning Outcomes:**

On successful completion of the course the student will be able to:
CLO1: Know about structure of chromosomes and chromatin model,
CLO2: Apply classical cytogenetic and molecular cytogenetic techniques for the identification of individual chromosomes and various chromosomal disorders,
CLO3: Describe biochemistry and genetics of various blood group types,
CLO4: Distinguish different disorders caused due to abnormal variants of hemoglobin, lysosomal enzymes, lipids and DNA nitrogenous bases.

| **UNIT I** **11 Hours** | **CLO1** |
|---|---|
| General features of Human Chromosome and Chromosome staining: Chromatin structure; nucleosome model; constitutive and facultative heterochromatin; centromeres, Telomere and its maintenance; nuclear organization region (NOR); chromosome nomenclature; sister chromatid exchanges (SCE); mosaicism; structure of human X and Y chromosome; ring chromosomes; human artificial chromosome. | |

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| **UNIT II** **12 Hours**<br>Cytogenetic and Molecular Cytogenetic Techniques: Methods of Chromosome preparation; chromosome banding techniques: G banding, Q banding, R banding and C banding; fluorescent in situ hybridization (FISH); different types of FISH probes: centromeric probes, chromosome specific probes and telomeric probes; reverse painting; flow cytometry; comparative genomic hybridization (CGH) mapping technique, whole chromosome painting; spectral karyotyping (SKY). | **CLO2** |
| **UNIT III** **11 Hours**<br>The concept of Biochemical Polymorphism: Concept of enzyme and protein polymorphism; molecular structure; Blood Group system: Biosynthesis and genetics of the ABH antigens; Rh antigens and MN antigens; quantitative and qualitative hemoglobin variants and its clinical affects | **CLO3** |
| **UNIT IV** **11 Hours**<br>Disorders due to abnormal variants of lysosomal enzymes, lipids and DNA nitrogenous bases: Disorders due to abnormal lysosomal enzymes: Tay-Sachs disease and Mucopolysaccharidoses; Disorders due to abnormal lipoprotein and lipid metabolism: hyper lipoproteinemia; Disorders due to abnormal purine metabolism: Lesch-Nyhan syndrome; disorders of pyrimidine metabolism: Orotic Aciduria. | **CLO4** |

**Transactional modes:** Lecture**;** Demonstration**;** Tutorial**;** Virtual classrooms; Lecture cum demonstration; Problem solving; Self-learning.

**Internal assessment shall be through any of the following:** Discussion on evolution of X and Y chromosomes, In Depth interview on banding techniques; Seminar on molecular cytogenetic techniques, Assignment on Flow cytometry; Surprise Tests, one sentence summary, Practice exercise on karyotype analysis, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, Seminars, term papers, discussions and presentations

**Suggested Readings:**

1. Gillham, N. (2011). Genes, Chromosomes and Disease. Pearson
2. Griffiths, A.J.F., Wessler, S.R. and Carroll, S.B. (2012). An Introduction to Genetic Analysis. W.H. Freeman Publication, USA.
3. Hein, S. and Mitelman, F. (2009). Cancer Cytogenetics. Wiley-Blackwell.
4. Korf, B.R and Irons, M.B. (2013). Human Genetics and Genomics. Wiley-Blackwell.
5. Kumar, A. and Srivastava, M. (2012) A textbook of Molecular Cytogentics, Narendra Publishing House, India
6. Purandare, H. and Chakravarty, A. (2000) Human Cytogenetic Techniques and Clinical Applications. Bhalani Publishing House, Mumbai, India.
7. Ram, M. (2010). Fundamental of Cytogenetics and Genetics. PHI Learning Pvt. Ltd.
8. Roy, D. (2009). Cytogenetics.Narosa Publishing House. New Delhi, India.
9. Tom, S and Read, A (2010). Human Molecular Genetics. Garland Science.
10. Shukla, A.N. (2009). Elements of enzymology. Discovery Publishing. New Delhi, India.
11. Voet, D. and Voet, J.G. (2008). Principles of Biochemistry. CBS Publishers & Distributors, New Delhi.

**Syllabus**
**M.Sc. in Human Genetics (Batch-2022)**

| | |
|---|---|
| **Course Code:** | **MME.522** |
| **Course Title:** | **Essentials of Immunology** |
| **Total Hours:** | **45** |

| L | T | P | C |
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| 3 | 0 | 0 | 3 |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:
CLO1: Evaluate basic concepts of immune system.
CLO2: Gain knowledge about various key processes related to development of immune system.
CLO3: Understand the concept of immune-based diseases as either a deficiency of components or excess activity as hypersensitivity.
CLO4: Apply the knowledge how immune system is involved in diseases caused by internal or external factors.

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| **Unit: 1** **12 Hours**<br>Immune System: The cells and organs of immune system, humoral immunity-immunoglobulin, basic structure, classes and subclasses, structural and functional relationships, nature of antigen, antigen-antibody reaction, antibody diversity, class switching, B and T cell development. | **CLO1** |
| **Unit: 2** **11 Hours**<br>Immune Effectors: Complement system, their structure, functions and mechanisms of activation by classical, alternative and lectin pathway. Th1 and Th2 response, various effector cells of immune system: DC, NK, Monocytes etc. | **CLO2** |
| **Unit: 3** **12 Hours**<br>Mechanisms of Immune System Diversity: Structure and functions of Major Histocompatibility Complex (MHC) and Human Leukocyte Antigen (HLA) system, polymorphism, distribution, variation and their functions. | **CLO3** |
| **Unit: 4** **10 Hours**<br>Immune System in Health and Diseases: Inflammation, hypersensitivity and autoimmunity, AIDS and immunodeficiencies, vaccine development. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Lecture cum demonstration; Problem solving; Self-learning.
**Internal assessment shall be through any of the following:** Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, presentations, and discussions.

**Suggested Reading:**

1. Kindt, T.J., Osborne, B.A. and Goldsby, R.A. (2018). Kuby Immunology. W.H. Freeman, USA.
2. Abbas. (2018). Cellular and Molecular Immunology.CBS Publishers & Distributors, India.
3. Charles, A. and Janeway, J.R. (2001). Immunobiology: The immune system in health and disease Blackwell Publishing, USA.
4. Delves, P.J., Roitt, I.M. and Seamus, J.M. (2016). Roitt's Essential Immunology (Series–Essentials). Blackwell Publishers, USA.

5. Elgert, K.D. (2015). Immunology: Understanding the immune system. Wiley-Blackwell, USA.
6. Tizard. (2018). Immunology: An Introduction. Cengage Learning, Thompson, USA.
7. Owen, Judith A; Punt, Jenni, Stranford, Sharon A. Kuby's Immunology (2013), W.H. Freeman and Company: New York, 2013

**Course Code:** **HGE.523**
**Course Title:** **Advanced Techniques in Human Genetics**
**Total Hours:** **45**

| L | T | P | C |
|---|---|---|---|
| 3 | 0 | 0 | 3 |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:
CLO1: Analyze various cellular processes using biochemical techniques.
CLO2: Learn the techniques for the visualization of a broad range of biological processes and features in cell structure.
CLO3: Learn the isolation of nucleic acids and its quantitative & qualitative analysis. Their importance for clinical molecular methods. Apply the knowledge to decipher the mechanisms of molecular and cell biology.
CLO4: Learn various immuno-techniques, Mutation analyses techniques and Cell and tissue culture techniques. Conceptualize principles of these different techniques used in life sciences.

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| **UNIT I** **11 Hours**<br>**Biochemical Techniques**: spectrometry; Mass, UV, IR, NMR and atomic absorption spectrophotometry; centrifugation: principle and applications; ultracentrifugation; | **CLO1** |
| **UNIT II** **11 Hours**<br>**Microscopy:** Light microscopy; phase contrast microscopy; fluorescent microscopy; scanning electron microscopy (SEM/FESEM); transmission electron microscopy (TEM); | **CLO2** |
| **UNIT III** **11 Hours**<br>**Nucleic acids:** Isolation, purification and analysis of nucleic acids; electrophoresis: principle of gel electrophoresis, polyacrylamide gel electrophoresis (PAGE and SDS-PAGE), agarose gel electrophoresis, pulse field gel electrophoresis (PFGE) and 2-Dimensional gel electrophoresis; polymerase chain reaction (PCR): principle, types and applications; PCR based markers:, SNPs; blotting techniques: southern, northern, western, dot blotting and hybridization; DNA fingerprinting. | **CLO3** |
| **UNIT IV** **11 Hours**<br>Flow Cytometry: Cell sorting; hybridoma technology: production of antibodies; histochemical and immuno techniques;; developing monoclonal and polyclonal antibodies; immunocytochemistry, radioimmunoassay (RIA); enzyme linked immunosorbent assay (ELISA) and autoradiography. Mutation analyses techniques: Restriction mapping, SSCP analyses, DNA sequencing-manual and automated methods. Cell and tissue culture techniques: Plants and animals. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Students visit to central instrument facility, Virtual classrooms; Lecture cum demonstration; Problem solving; Self-learning.

**Internal assessment shall be through any of the following:** Interview on various microscopic techniques; Visit to CIL and compile a report; Case studies solved by DNA fingerprinting, Assignment; In Depth interviews, Analysis of DNA sequencing data, Surprise Tests, term paper, Seminars, discussions and presentations, drawing flow charts for the techniques.

**Suggested Readings:**

1. Brown, T.A. (2010). Gene cloning and DNA analysis: An Introduction. 6th Edition, Wiley-Blackwell Publisher, New York.
2. Goldsby, R.A., Kindt, T.J. and Osborne, B.A. (2008). Kuby Immunology. 6th Edition, W. H. Freeman & Company, San Francisco.
3. Gupta, P.K. (2005). Elements of biotechnology. Rastogi Publications, Meerut.
4. Gupta, S. (2005). Research methodology and statistical techniques, Deep & Deep Publications (P) Ltd. New Delhi.
5. Kothari, C.R. (2008.) Research methodology(s). New Age International (P) Ltd., New Delhi
6. Lewin, B. (2010). Genes X, CBS Publishers & Distributors. New Delhi.
7. Mangal, S.K. (2007). DNA Markers in Plant Improvement. Daya Publishing House, New Delhi.
8. Nelson, D. and Cox, M.M. (2009). Lehninger Principles of Biochemistry. W.H. Freeman and Company, New York.
9. Primrose. S.B. and Twyman, R. (2006). Principles of Gene Manipulation and Genomics. Blackwell Publishing Professional, U.K.
10. Sambrook, J. (2006). The Condensed Protocols from Molecular Cloning: A Laboratory Manual .Cshl Press. New York.
11. Sambrook, J. and Russell, D.W. (2000). Molecular Cloning: A Laboratory Manual (3 Vol-set). 3rd Edition, CSHL Press, New York.
12. Sawhney, S.K. and Singh, R. (2005). Introductory Practical Biochemistry. Narosa Publishing House, New Delhi.
13. Slater, A., Scott, N.W., and Fowler, M.R. (2008). Plant Biotechnology: The Genetic Manipulation of Plants. Oxford University Press, USA.
14. Wilson, K. and Walker, J. (2006). Principles and Techniques of Biochemistry and Molecular biology. 6th Edition, Cambridge University Press India Pvt. Ltd., New Delhi.
15. Sue Carson Heather Miller Melissa Srougi D. Scott Witherow (2019) Molecular Biology Techniques. Academic Press, USA

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|---|---|
| **Course Code:** | **HGE.524** |
| **Course Title:** | **Human Anatomy and Physiology** |
| **Total Hours:** | **45** |

| L | T | P | C |
|---|---|---|---|
| 3 | 0 | 0 | 3 |

**Syllabus**
**M.Sc. in Human Genetics (Batch-2022)**

**Learning outcome**

CLO1: Understand anatomical and physiological functions of various tissues.
CLO2: Understand the clinical scenarios and be able to interpret physiological function.
CLO3: Recognize the cell structure and function, histology, gross anatomy and physiology of several organ systems.
CLO4: Understand and predict the body response to stimuli. Recognize the principle of homeostasis and control mechanisms

| | |
|---|---|
| **UNIT I** **11 Hours**<br>Muscular System: Structure and organization of muscles: skeletal, cardiac and smooth muscles; neuromuscular junction.<br>Cardiovascular System: Physiological anatomy of Heart; cardiac muscle, cardiac cycle; blood constituents; hematopoiesis; cardiovascular regulation. | **CLO1** |
| **UNIT II** **12 Hours**<br>Digestive System: Anatomy of Gastrointestinal tract, physiology of salivary secretion, mouth and esophagus, epithelial barrier function; digestion and absorption in GIT; GIT secretions and accessory glands; BMR.<br>Urinary System: Physiological anatomy Kidney; urine formation; regulation of volume and concentration of body fluids, KFT. | **CLO2** |
| **UNIT III** **11 Hours**<br>Nervous System: Organization of nervous system, synapse, generation of action potential; vision; hearing and tactile response, degeneration and regeneration of peripheral nerves.<br>Thermoregulation and stress adaptation: Comfort zone; body temperature – physical, chemical and neural regulation; acclimatization. | **CLO3** |
| **UNIT IV** **11 Hours**<br>**Respiratory System**: Anatomical considerations; mechanism of respiration; neural and chemical regulation of respiration; Physiology of high altitude, hypoxia, PFT.<br>**Reproduction**: Physiology of reproductive system (male, female), pregnancy, physiology of fetus. | **CLO4** |

**Transactional Modes:** Lecture**;** Seminar**;** Tutorial**;** Virtual classrooms; Problem solving; Self-learning; group discussion.

**Internal assessment shall be through any of the following:** Assignment; In Depth interviews, Surprise Tests, term paper, Seminars, discussions, and presentations.

**Suggested Readings:**


1. Brody, T. (1998). Nutritional biochemistry. Academic Press, USA.
2. Devlin, T.M. (2005). Textbook of Biochemistry with clinical correlations. John
2. Wiley & Sons Inc. USA.
3. Guyton. (2007). Textbook of medical physiology. 11th Edition. Elsevier India Pvt. Ltd. New Delhi.
4. Hill, R.W, Wyse, G. A. and Anderson, M. (2008). Animal physiology. Sinauer Associates Inc. USA.
5. Khurana. (2006). Textbook of medical physiology. Elsevier India Pvt. Ltd.
6. Murray, R.K. (2009). Harper's illustrated biochemistry. Jaypee Publishers,

7. New Delhi, India.
8. Tyagi, P. (2009). A textbook of Animal Physiology. Dominant Publishers and
9. distributors, New Delhi, India.
10. Silverthorne D, (2011) Human Physiology, Pearson; 6th edition.
11. Sherman V. (2013) Vander's Human Physiology. McGraw-Hill 13th edition.
12. Jain A.K. (2021). Textbook of physiology. Avichal Publishing Company-9th Edition.

| | | L | T | P | C |
|---|---|---|---|---|---|
| **Course Code:** | **MME.525** | **0** | **0** | **2** | **1** |
| **Course Title:** | **Essentials of Immunology (Practical)** | | | | |
| **Total Hours:** | **30** | | | | |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:
CLO1: Understand the basic concepts in handling of blood samples.
CLO2: Identification of different cells in blood
CLO3: Conceptualize the Immunologic techniques used in diagnosis of diseases.
CLO4: Hands on experience in purification of antibodies.

List of Practicals

| | | |
|---|---|---|
| 1. | Separation of Plasma and Serum from the blood samples. | **CLO1** |
| 2. | Blood film preparation and identification of cells | **CLO2** |
| 3. | Separation of mononuclear cells | |
| 4. | Lymphoid organs and their microscopic organization | |
| 5. | Double diffusion and immuno-electrophoresis | **CLO3** |
| 6. | ELISA | |
| 7. | Radial immuno diffusion | |
| 8. | Purification of IgG from serum | **CLO4** |

*Practical can be modified depending upon the available faculty/facility.

**Suggested Reading:**

1. Practical immunology (2002) by F.C. Hay and O.M.R. Westwood, P.N. Nelson, L. Hudson (Wiley-Blackwell).
2. Clinical immunology and serology: A laboratory perspective (1997) by Stevens C.D (FA Davis Company, Philadelphia).

**Transactional Modes:** Hands-on Practicals; Demonstration**;** Tutorial**;** Self-learning.

**Evaluation criteria:**

A. Continuous Assessment = **60 Marks**
Based on performance and good lab practices of the students and completion of the record book

(60 divided by the number of practicals)

B. Final Examination = **40 Marks**

ii. Subjective question = 10 Marks
iii. Performing experiment = 20 Marks
iv. Viva voce = 10 Marks

**Course Code:** **HGE.526**
**Course Title:** **Clinical Genetics (Practical)**
**Total Hours:** **45**

| L | T | P | C |
|---|---|---|---|
| 0 | 0 | 6 | 3 |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:
CLO1: Interpret how genetic factors predispose to mendelian and adult onset diseases
CLO2: Implications of these predispositions for diagnosis, prevention, and treatment.
CLO3: Appreciate the importance of genetic counseling to assert the birth of a children affected with genetic disorders
CLO4: Assess the clinical features of common chromosomal alterations.
CLO5: Gain an insight into specific exposures that are likely to be teratogenic in humans
CLO6: Application of appropriate molecular biology techniques for diagnosis, disease prediction and prevention.

**List of Practical work:**

| | | |
|---|---|---|
| 1 | Genetic assessment and drawing pedigree. | **CLO1** |
| 2 | Genetic counseling | **CLO2, CLO3** |
| 3 | Common chromosomal disorders-case studies | **CLO4** |
| 4 | Teratogens and dysmorphology | **CLO5** |
| 5 | DNA and RNA isolation, quantification, gel electrophoresis (agarose/PAGE), | **CLO6** |
| 6 | PCR-RFLP | |
| 7 | RT-PCR | |
| 8 | ELISA | |

**Transactional Modes:** Lecture**;** Demonstration**;** Virtual classrooms; Problem solving; Self-learning, Pedigree drawing.

**Evaluation criteria for practical courses:**
- Continuous Assessment = **60 Marks**

Based on performance and good lab practices of the students and completion of the record book (60 divided by the number of practicals)
- Final Examination = **40 Marks**

  Subjective question = 10 Marks
  Performing experiment = 20 Marks
  Viva voce = 10 Marks

**Suggested Readings:**

1. Brooker, R.J (2017). Genetics: analysis and principles. 6th Ed. New York, NY: McGraw-Hill Education.
2. Hartwell, L. et al (2017). Genetics: from genes to genomes. 6th Ed. New Your, NY: McGraw-Hill Education.
3. Helen, M.K. 2002. ABC of clinical Genetics. 3rd Ed. BMJ Publishing Group. London.

**Course Code:** **MME.528**
**Course Title:** **Molecular Biology**
**Total Hours:** **45**

| L | T | P | C |
|---|---|---|---|
| 3 | 0 | 0 | 3 |

**Course Learning Outcomes:**

On successful completion of the course the student will be able to:

CLO1: Conceptualization of molecular mechanisms involved in cellular functioning.
CLO2: Understanding the molecular processes of DNA replication
CLO3: Study the process of transcription in prokaryotes and eukaryotes.
CLO4: Understand the concept of Genetic code, process of translation and post translational modifications

| | |
|---|---|
| **Unit: 1** **12 Hours**<br>Nucleic acids, Genes and Genome organization: Chemical structure of DNA and base composition, Watson-Crick model, mitochondrial DNA, Chromosome Structure, Chromatin and the Nucleosome, Chromatin structure: euchromatin, heterochromatin, Constitutive and facultative heterochromatin, Regulation of chromatin structure and nucleosome assembly, typical structure of a eukaryotic genes including various regulatory elements. | **CLO1** |
| **Unit:2** **12 Hours**<br>DNA replication and repair: Mechanisms of DNA replication in eukaryotes, Enzymes and accessory proteins involved in DNA replication, Replication errors and proofreading, telomeres, DNA damage and repair mechanisms. | **CLO2** |
| **Unit: 3** **11 Hours**<br>Transcription and mRNA processing: Different forms of RNA: mRNA, tRNA, rRNA and other Types of RNA Eukaryotic transcription: Initiation, Elongation & Termination, general and specific transcription factors, Regulatory elements and mechanisms of transcription regulation, RNA processing and editing, post transcriptional gene regulation. | **CLO3** |
| **Unit: 4** **10 Hours**<br>Translation: Genetic code, eukaryotic translation, the translation machinery, mechanisms of chain initiation, elongation and termination, regulation of translation, co-and post- translational modifications of proteins. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Lecture cum demonstration; Problem solving; Self-learning.

**Internal assessment shall be through any of the following:** Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, presentations and discussions.

**Suggested Reading:**

9. Sambrook, J., Fritsch, E. F., & Maniatis, T. (2015). Molecular cloning: a laboratory manual. Cold Spring Harbor Laboratory Pressn New York.
10. Berk, A. Chris, A.K. & Krieger, M. (2011). Molecular Cell Biology. W.H. Freeman, USA.
11. Robertis, (2011). Cell and Molecular Biology. Lippincott Williams & Wilkins.
12. Karp, G. (2010). Cell and molecular biology: concepts and experiments. John Wiley & Sons.
13. Krebs, J. E., Goldstein, E. S., & Kilpatrick, S. T. (2017). Lewin's Genes XII. Jones & Bartlett Learning.
14. Watson, J. D., Baker, T. A., Bell, S. P., Gann, A., & Levine, M. (2003). Molecular Biology of the Gene Benjamin Cummings.
15. Alberts, B., Johnson, A., Lewis, J., Raff, M., Roberts, K., & Walter, P. (2007). Molecular biology of the cell. Garland Science. New York, 1392.
16. Fasman, G.D. (1989). Practical Handbook of Biochemistry and Molecular Biology. CRC Press, Taylor and Francis Group, UK.

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|---|---|
| **Course Code:** | **HGE.529** |
| **Course Title:** | **Principles of Ecological Science** |
| **Total Hours:** | **30** |

| L | T | P | C |
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**Course Learning Outcomes:**

On successful completion of the course the student will be able to:

CLO1: Improve their knowledgebase about basics of ecological science

CLO2: Conceptualize and contribute in environmental studies

CLO3: Improve student's aptitude for research and development on ecological succession and dynamics

CLO4: Contribute to conservation science

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| **UNIT I** **6 Hours**<br>**Environmental components:** Physical environment; biotic environment; biotic and abiotic interactions. Concept of habitat and niche; resource partitioning; character displacement. Major terrestrial biomes; theory of island biogeography; biogeographical zones of India. | **CLO1** |
| **UNIT II** **8 Hours**<br>**Biological components of environment:** Characteristics of a population; life history strategies (r and K selection); concept of metapopulation – demes and dispersal, interdemic extinctions, age structured populations; Species Interactions: Types of interactions, interspecific competition, herbivory, carnivory, pollination, symbiosis; | **CLO2** |

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| Community Ecology: Nature of communities; community structure and attributes; levels of species diversity and its measurement; edges and ecotones. | |
| **UNIT III** **8 Hours**<br>**Ecosystem and Ecological Succession:** Ecosystem: structure and function; energy flow and mineral cycling (C, N, P); primary production and decomposition; structure and function of some Indian ecosystems: terrestrial and aquatic. Ecological Successions: Types; mechanisms; changes involved in succession; concept of climax. | **CLO3** |
| **UNIT IV** **8 Hours**<br>**Applied Ecology and Conservation Biology:** Environmental pollution; biodiversity: status, monitoring and documentation; biodiversity management approaches; Principles of conservation and its management; Indian case studies on conservation/management strategy: Project Tiger, Biosphere reserves. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Lecture cum demonstration; Problem solving; Self-learning; MCQ practice.

**Internal assessment shall be through any of the following:** Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, Seminars, term papers, discussions, and presentations.

**Suggested Readings:**

1. Smith, Robert Leo, et al. "Elements of ecology." (2015): E9.
2. Recknagel F. 2002 Ecological Informatics: Understanding Ecology by Biologically-Inspired Computation, Springer, New York.
3. Odum E.P. 1983 Basic Ecology. Saunders International Edition, Japan
4. Michael Begon, 2020 Ecology: From Individuals to Ecosystems 5th Edition,

## Discipline Elective -II

**Course Code:** **HGE.527**
**Course Title:** **Human Embryology and Developmental Genetics**
**Total Hours:** **45**

| L | T | P | C |
|---|---|---|---|
| 3 | 0 | 0 | 3 |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:
CLO1: Conceptualize basics of reproductive physiology
CLO2: Correlate genetic regulation in different embryonic developmental stages
CLO3: Evaluate the role of biomolecules in embryonic development.
CLO4: Know different genetic and environmental triggers for post-natal development, ageing and senescence.

**Syllabus**
**M.Sc. in Human Genetics (Batch-2022)**

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|---|---|
| **UNIT I** 12 Hours<br>Basic concepts of development: Gametogenesis; fertilization; embryogenesis: gastrulation and implantation of embryo; fetal membrane and placenta; potency, commitment, specification, induction, competence, determination, and differentiation. Role of Sry, Sox9 and WNT4 and DAX1 in early gonad differentiation | **CLO1** |
| **UNIT II** 11 Hours<br>Gene expression regulation in development: Basics of gene expression regulation during early embryogenesis; homeotic genes, P granules, role of key developmental genes: polycomb gene, SOX, HOX. | **CLO2** |
| **UNIT III** 11 Hours<br>Stem Cell and Organogenesis: Stem cell: embryonic and adult; cell-cell communication; neural crest cells and axonal specificity; vertebrate eye and central nervous system development; hematopoiesis. | **CLO3** |
| **UNIT IV** 11 Hours<br>Post-natal Development, Aging and senescence: Environmental and genetic factors during maturations, Sex linked changes, Deciduous and primary teeth, Clinical death. Teratology: Teratogens, introduction to toxicogenomic. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Virtual classrooms; Lecture cum demonstration; Problem solving; Self-learning.
**Internal assessment shall be through any of the following:** Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, Seminars, term papers, discussions, and presentations.

**Suggested Readings:**


1. Gilbert, S.F. (2013). Developmental Biology. Tenth Edition.
2. Slack, J.M.W. (2012). Essentiel Developmental Biology. Third Edition.
3. Moody, A.A. (2014). Principles of Developmental Genetics. Second Edition.
4. Slack, J.M.W. (2018). The Science of Stem Cells. First Edition.
5. Milunsky, J. & Milunsky, A. (2010). Genetic Disorders and the Fetus: Diagnosis, Prevention & Treatment. Willey Blackwell India, New Delhi.
6. Prakash, G. (2007). Reproductive Biology. Narosa Publication House Pvt. Ltd., New Delhi.
7. Sadler, T.W., Tosney, K., Chescheir, N.,C., Imseis, H., Leland, J. and Sadler-Redmond, S.,L. (2011).Langman's Medical Embryology (Longmans Medical Embryolgy). Lippincott Williams and Wilkins.
8. Keith L. Moore BA, T. V. N. Persaud MD., Mark G. Torchia (2019 The Developing Human Clinically Oriented Embryology, Elsevier, Netherlands


**Course Code:** **MME.527**
**Course Title:** **Stem Cell and Regenerative Medicine**
**Total Hours:** **45**

| L | T | P | C |
|---|---|---|---|
| 3 | 0 | 0 | 3 |

**Course Learning Outcomes:**

On successful completion of the course the student will be able to:
CLO1: Understand basics of cell culture
CLO2: Understand basic stem cell biology.
CLO3: Gain conceptual knowledge about requirements for tissue engineering.
CLO4: Know regenerative medicine and its potential applications.

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| **Unit: 1** **11 Hours**<br>Basics of cell culture and media, Culturing primary cells and cell lines, suspension and adherent cultures, cell growth, growth inhibition and apoptotic studies, Embryo culture, transplantation and teratogens, teratomas, stem cell culture, organ culture. | **CLO1** |
| **Unit: 2** **12 Hours**<br>Stem Cells: Stem cells and their properties, classification of stem cells, in-vitro culture techniques, isolation, identification and characterization of stem cells, stem cells in various organs and in disease conditions. | **CLO2** |
| **Unit: 3** **11 Hours**<br>Tissue Engineering: Principles of tissue culture, tissue and organ culture, extracellular matrices, bioreactors, ethical issues related to stem cell therapies, stem cell banks, bone marrow transplantation. | **CLO3** |
| **Unit: 4** **11 Hours**<br>Regenerative Medicine: Modes of tissue and organ delivery, tissue Engineering and transplantation techniques, immuno isolation techniques, regeneration of bone and cartilage, Islet cell transplantation and bio-artificial pancreas, lung regeneration | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Lecture cum demonstration; Problem solving; Self-learning.
**Internal assessment shall be through any of the following:** Group discussions, Surprise Tests, case analysis, simulated problem solving, classroom assignments, term paper, presentations and discussions.

**Suggested Reading:**

1. Lanza, R., Gearhart,J. (2016). Essential of Stem Cell Biology. Elsevier Academic Press.
2. Lanza, R., Klimanskaya, I. (2018). Essential Stem Cells Methods. Academic Press.
3. Mao, J. J. & (2017). Translational approaches in tissue engineering and regenerative medicine. Artech House.
4. Lanza, R. (2017). Principles of Tissue Engineering, 3rd Edition. Academic Press
5. Stein, G. S., Borowski, M., Luong, M. X., Shi, M. J., Smith, K. P., & Vazquez, P. (Eds.). (2011). Human stem cell technology and biology: A research guide and laboratory manual. John Wiley & Sons.
6. Lanza, R., Blau, H., Gearhart, J., Hogan, B., Melton, D., Moore, M., ... & Weissman, I. (Eds.). (2014). Handbook of Stem Cells, Two-Volume Set: Volume 1-Embryonic Stem Cells; Volume 2-Adult & Fetal Stem Cells. Elsevier.

## Semester – III

**Syllabus**
**M.Sc. in Human Genetics (Batch-2022)**

| | |
|---|---|
| **Course Code:** | **HGE.551** |
| **Course Title:** | **Pharmacogenomics and Nutrigenomics** |
| **Total Hours:** | **45** |

| L | T | P | C |
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**Course Learning Outcomes:**
On successful completion of the course the student will be able to:
CLO1: Define pharmacogenomics and will understand its application in clinical setting. The students will be aware about the challenges in the field on account of different ethnic background.
CLO2: Have an understanding about the nutritional requirements and the role of gut microbiota in human nutrition.
CLO3: Be aware of the effect of genetic polymorphisms in variable response to micronutrients and will understand the regulation of transcription factors by the nutrients.
CLO4: Get aware of the genetic markers involved in the regulation of metabolomics and the role of these markers in health and disease.

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| **UNIT I** **12** Hours<br>Pharmacogenomics: Pharmacokinetics and pharmacodynamics; drug-metabolizing enzymes: cytochrome P450s, VKORC1 and TPMT; personalized treatment: example of warfarin, anti-epileptic and anti-cancer drugs like methotrexate and tamoxifen, trastuzumap; heredity disorders with altered drug response: porphrriavariegata, hemoglobinopathies, Grigler-najjar syndrome; concept of pharmacogenomics; | **CLO1** |
| **UNIT II** **11** Hours<br>Nutritional Biochemistry: Essential and non-essential nutrients; micro and macro nutrients; basal metabolic rate (BMR); malnutrition; malabsorption and interventional strategies. Concept of gut microbiota in human nutrition.<br>Gut-brain axis in absorption; Role of Prebiotics and Probiotics in Gut brain axis | **CLO2** |
| **UNIT III** **11** Hours<br>Nutrigenomics in determining health: Diet and gene expression; nutritional status in early life and metabolic programming; nutrients as regulators of activity and transcription factors; modulating the risk of obesity and vitamin D deficiency through nutrigenomics. | **CLO3** |
| **UNIT IV** **12** Hours<br>Biomarkers and recent advances in Nutrigenomics: Genetic and nutritional control of lipid metabolism, metabolomics; effect of diet on epigenetic processes, concept of oral and gut microbiome and its implications in health and disease; therapeutic approaches through microbiota transplantation. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Virtual classrooms; Lecture cum demonstration; Problem solving; Self-learning.
**Internal assessment shall be through any of the following:** Literature survey on current medicines based on pharmacogenomics, In-depth interview on ADRs. Group activity to calculate BMI, Preparation of a list of essential micro and macronutrients, Surprise Tests, case analysis,

classroom assignments, homework assignments, term paper, Seminars, term papers, discussions, and presentations.

**Suggested Readings:**

1. Altman RB, Flockhart D and Goldstein DB (2012). Principles of Pharmacogenetics and Pharmacogenomics. Cambridge University Press.
2. Ferguson, L,R.(2013) Nutrigenomics and Nutrigenetics in Functional Foods and Personalized Nutrition. CRC Press.
3. Tollefsbol T (2011). The New Molecular and Medical Genetics. Elsevier Inc
4. Simopoulos A.P. and Ordovas J.M. (2004). Nutrigenetics and Nutrigenomics Karger Publishers
5. Rimbach, G and Fuchs, J (2005) Nutrigenomics (Oxidative Stress and Disease). CRC press
6. Yui-Wing Francis Lam Stuart Scott (2018) Pharmacogenomics: Challenges and Opportunities in Therapeutic Implementation, Elsevier, Netherlands

**Course Code:** **HGE.552**
**Course Title:** **Genetic Diseases and Therapies**
**Total Hours:** **45**

| L | T | P | C |
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**Course Learning Outcomes:**

On successful completion of the course the student will be able to:
CLO1: Understand the genetic causes of monogenic disorders and common multifactorial diseases
CLO2: Understand the genetic basis of Genomic imprinting. Analyze the consequences of altered epigenetic processes in causing genetic disorders
CLO3: Understanding genetic screening
CLO4: Apply the principal of genetics and biostatistics for genetic disorders risk assessment

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| **UNIT I** **11** Hours<br>Monogenic Disorders: Cystic fibrosis; Huntington's disease; Duchenne Muscular dystrophy; X-linked rickets. Multifactorial Diseases: Diabetes type 2; Cancers; Hypertension; Obesity; Neurodegenerative diseases, Depression, IBS | **CLO1** |
| **UNIT II** **11** Hours<br>Genomic Imprinting and Human Diseases: Uniparental disomy & genomic imprinting: Syndromes: Prader-Willi & Angelman syndrome; Beckwith-Wiedemann syndrome & Silver Russell Syndrome; Role of Imprinting brain development and behavior. | **CLO2** |
| **UNIT III** **11** Hours<br>Genetic Screening: Pre symptomatic testing for genetic diseases and malignancy, carrier detection; prenatal and postnatal screening; Assisted reproductive techniques and Pre-implantation diagnosis; Genetic Counseling. | **CLO3** |

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| **UNIT IV** **12** Hours<br>Therapies for genetic disorders and multifactorial diseases: Stem cell therapies: stem cell types, cord blood cells, bone marrow transplantation; current stem cell therapies; gene therapies: methods; diseases suitable for gene therapies: hemoglobinopathies, cystic fibrosis,<br>muscular dystrophies, cancer; challenges in gene therapy; regulatory requirements, RNA Therapies | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Virtual classrooms; Lecture cum demonstration; Problem solving; Self-learning.

**Internal assessment shall be through any of the following:**

Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, Seminars, term papers, discussions and presentations, Assignment on various gene therapies in use currently, Group activity to explore regulatory requirements and prepare a report.

**Suggested Readings:**

1. Brown, S.M., (2009). Essentials of Medical Genomics. Wiley-Blackwell.
2. Jocelyn, E. K., Elliot, S. G., Stephen, T. K. (2009), Lewin's Gene X. Jones &Barlett.
3. Krebs, J.E., Goldstein, E.S. and Kilpatrick, S.T. (2014). Lewin's Genes XI. Jones and Bartlet India Pvt. Ltd.
4. Lodish, H., Berk, A., Chris, A. K., Krieger, M. (2008), Molecular Cell Biology. W.H. Freeman, USA.

**Course Code:** **HGE.553**
**Course Title:** **Recombinant DNA Technology and Therapeutics**
**Total Hours:** **45**

| L | T | P | C |
|---|---|---|---|
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**Course Learning Outcomes:**

On successful completion of the course the student will be able to:
CLO1: Demonstrate cloning and expression vectors in E.Coli in recombinant DNA techniques
CLO2: Get familiar with vectors for Eukaryotes
CLO3: Learn conventional methods used in recombinant DNA technology
CLO4: Learn therapeutics aspect of recombinant DNA technologies

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| **UNIT I** **11 Hours**<br>Basics of Genetic Engineering: Isolation and purification of nucleic acids; restriction and DNA modifying enzymes; restriction modification systems; cloning vectors: plasmids, phages, lambda vectors, cosmids, PAC, BAC, YAC; selection and screening of clones. | **CLO1** |
| **UNIT II** **11 Hours**<br>Construction of DNA libraries: Genomic and cDNA libraries; screening of genomic and expression libraries. | **CLO2** |

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| Analysis of gene expression: Real-time-PCR; digital PCR; RNA sequencing analysis; EST analysis; promoter analysis; TSS mapping; cDNA and oligos array; serial analysis of gene expression (SAGE). | |
| **UNIT III** **11 Hours**<br>Expression Vectors: Animal virus derived vectors-Sv-40, caccinal/bacculo, retroviral vectors and pichia vectors system; Expression vectors and Protein purification methods, Inclusion bodies and methodologies to reduce its formation. | **CLO3** |
| **UNIT IV** **12 Hours**<br>Techniques and Applications of recombinant DNA technology: Sites specific mutagenesis; Gene Editing through; CRISPR/CAS system, Genetically modified microbes (Recombinant bacteria) for the production of commercial scale production of proteins, pharmaceuticals; transgenic animals; RNA based therapeutics. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Virtual classrooms; Lecture cum demonstration; Problem solving; Self-learning.

**Internal assessment shall be through any of the following:** Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, home work assignments, term paper, Seminars, term papers, discussions and presentations

**Suggested Readings:**

1. R.W. Old & S.B. Primrose (2007) Principles of Gene Manipulation 7th Edition Blackwell science.
2. Bernard R. Glick & Jack J. Pasternak. (2010) Molecular Biotechnology 4th Edition ASM Press Washington.
3. James Watson, Micheal Gilman Jan Witkowsk (2007) Recombinant DNA 3rdedition . CSHL, New York.
4. CokinRateldge and Bjorn Christiansen, (2006) Basic Biotechnology 3rdEdition Cambridge University press.
5. John E. Smith. (2009) Biotechnology 5th Edition by Cambridge University press.
6. Molecular Biology of Gene 6th Edition by Watson CSHL Press New York.
7. Sambrook& Russell Molecular cloning , CSHL Press, New York.
8. David & Freifelder John & Barlett (2008) Molecular biology 2nd Edition, Narosa publishing, New Delhi.

**Related Weblinks:**

1. http://www.genengnews.com/ontheweb.asp
2. http://www.ige-india.com/
3. http://www.icgeb.org/~bsafesrv/
4. http://www.livescience.com/32648-whats-genetic-engineering.html

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| **Course Code:** | **MME.554** |
| **Course Title:** | **Tools of Bioinformatics (Practical)** |
| **Total Hours:** | **60** |

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**Course Learning Outcomes:**

On successful completion of the course the student will be able to:

1. Analyze publicly available and experimental genomics data
2. Perform in-silico prediction of protein structures and interactions
3. Perform in-silico functional annotation of genetic findings
4. Demonstrate physical property of biomolecules in silico

1. Access to sequence databases (downloading DNA/RNA/Protein sequences).
2. Performing sequence alignment using various tools (BLAST, MSA)
3. Pipeline of RNA-seq analysis.
4. Submission of SRA and TSA database.
5. Genome wide association study and DNA microarray-data analysis.
6. Understanding the evolutionary relationship using molecular phylogeny analysis.
7. Prediction of Protein structure using sequence database
8. Practical insights of tertiary structure prediction and comparative modelling
9. Protein-protein and protein-ligand docking
10. Evaluation of techniques for 3-D structure determination like X-ray, NMR, MS/MS analysis using case study.
11. Structure-based drug design and virtual screening of the drug.
12. Quantitative structure activity relations, Cheminformatics and pharmacophore mapping in therapeutic development.

**Transactional Modes:** Hands-on training; Demonstration**;** Tutorial**;** Lecture cum demonstration; Problem solving; Self-learning.
**Internal assessment shall be through any of the following:** Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, Seminars, term papers, discussions, and presentations.

**Evaluation criteria for practical courses:**

- Continuous Assessment = **60 Marks**

Based on performance and good lab practices of the students and completion of the record book (60 divided by the number of practicals)

- Final Examination = **40 Marks**

  Subjective question = 10 Marks
  Performing experiment = 20 Marks
  Viva voce = 10 Marks

**Suggested Readings:**

1. Laboratory Protocols. Third Edition. 2005. CIMMYT Applied Molecular Genetics Laboratory.
2. Koliantz, G & Szymanski, D.B. Genetics A Laboratory Manual. Second Edition. 2009. American Society of Agronomy, Crop Science Society of America.
3. Sambrook, J & Green, M.R. Molecular Cloning: A laboratory manual. Fourth Edition. 2013. Cold Spring Harbor Laboratory Press, U.S.
4. Pazos, F & Chagoyen, M. Practical protein bioinformatics.2015. Springer International Publishing, Switserland.

5. Agostino, M. Practical Bioinformatics. 2012. Garland Science. Taylor & Francis Group. New York and London.

# Discipline Elective-III

| | | L | T | P | C |
|---|---|---|---|---|---|
| **Course Code:** | **HGE.555** | | | | |
| **Course Title:** | **Biosafety, Bioethics, and Intellectual Property Rights** | **3** | **0** | **0** | **3** |
| **Total Hours:** | **45** | | | | |

**Course Learning Outcomes:**

On successful completion of the course the student will be able to:

CLO1: Interpret the bioethical issues concerning biotechnological advancements like recombinant DNA technology, cloning, gene manipulation.

CLO2: Implement biosafety while carrying out research.

CLO3: Distinguish different types of Intellectual Property Rights.

CLO4: Describe the ways of protecting traditional knowledge from Biopiracy.

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| **UNIT I** **11 Hours**<br>Biosafety: Good laboratory practices; Risk and safety assessment from genetically engineered organisms; special procedures for r-DNA based products; biological containment (BC) and physical containment (PC); CDC biosafety levels; biohazard management. | **CLO1** |
| **UNIT II** **11 Hours**<br>Bioethics: Ethical considerations during research, Use of Animals for clinical research, Embryonic and adult stem cell research, assisted reproductive technologies, cloning, MTP and Euthanasia; the element of informed consent; ethical issues of the human genome project. | **CLO2** |
| **UNIT III** **11 Hours**<br>Intellectual Property Rights (IPRs): Various forms of IP – patents, industrial designs, trademark, geographical indications, and plant breeder's right; copyright: fair use, plagiarism; protection of indigenous intellectual property. | **CLO3** |
| **UNIT IV** **12 Hours**<br>Patent system: Patent filing procedure in India and ways of patent protection in other countries: Determination of patentability of inventions, filing a patent application in India: timeline, procedure involved in the granting of a patent, patent cooperation Treaty (PCT). | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Virtual classrooms; Tutorial**;** Lecture cum demonstration; Problem solving; Case study; Self-learning.

**Internal assessment shall be through any of the following:**

Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, Seminars, term papers, discussions, and presentations.

**Suggested Readings:**

1. Clarke, A (2012). Genetic Counseling: Practice and Principles. Taylor & Francis
2. Fleming, D.O. and Hunt, D.L. (2006). Biological Safety: Principles and Practices. American Society for Microbiology, USA.
3. Mahop, M.T. (2010). Intellectual Property, Community Rights and Human Rights: The Biological and Genetic Resources of Developing Countries. Routledge.
4. Shannon, T.A. (2009). An Introduction to Bioethics. Paulist Press, USA.
5. Thompson J and Schaefer, B.D (2013).Medical Genetics: An Integrated Approach. McGraw Hill.
6. Vaughn, L. (2009). Bioethics: Principles, Issues, and Cases. Oxford University Press, UK.
7. WHO. (2005). Laboratory Biosafety Manual. World Health Organization.
8. Ahuja, V.K. (2017). Law relating to Intellectual Property Rights. LexisNexis, India. 3rd Edition.
9. Mahop, M.T. (2010). Intellectual Property, Community Rights and Human Rights: The Biological and Genetic Resources of Developing Countries. Routledge, USA.
10. Neeraj, P. and Khusdeep, D. (2014). Intellectual Property Rights. India, IN: PHI learning Private Limited.
11. Nithyananda, K V. (2019). Intellectual Property Rights: Protection and Management. India, IN: Cengage Learning India Private Limited.

**Course Code:** **MME.555**
**Course Title:** **Evolution and Developmental Biology**
**Total Hours:** **45**

| L | T | P | C |
|---|---|---|---|
| 3 | 0 | 0 | 3 |

**Course Learning Outcomes:**

On successful completion of the course the student will be able to:
CLO1: Gain knowledge on concept related to origin of life.
CLO2: Develop understanding regarding molecular evolution and original of first cell.
CLO3: Know about concepts related to developmental processes.
CLO4: Understand pathology related to mechanisms of development and differentiation.

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| **Unit: 1** **11 Hours**<br>**Origin of Life:** History of evolutionary ideas, Modern evolution theory, Natural Selection, Adaptation, The origin of species. | **CLO1** |
| **Unit: 2** **11 Hours**<br>**Paleontology and Molecular Evolution:** The evolutionary time scale, Major events in the evolutionary time scale, Intimate partnership, Stages in primate evolution, Human evolution, Neutral evolution, Molecular divergence and molecular clocks, Gene duplication and divergence. | **CLO2** |

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| **Unit: 3** **12 Hours**<br>**Basic Concepts of Development:** Potency, Commitment, Specification, Induction, Competence, Determination and Differentiation, Morphogenetic gradients, Cell fate and cell lineages, Stem cells, Genomic equivalence and the cytoplasmic determinants, Imprinting, Mutants and transgenics in analysis of development. | **CLO3** |
| **Unit: 4** **11 Hours**<br>**Gametogenesis, Fertilization, embryology and neurulation:** Production of gametes, Cell surface molecules in sperm-egg recognition; embryonic development and formation of germ layers in humans, fetal development, sex determination, neural tube formation. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Lecture cum demonstration; Problem solving; Self-learning.

**Internal assessment shall be through any of the following:** Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, Seminars, term papers, discussions, and presentations.

**Suggested Reading:**

1. Darwin, C. (1956). The Origin of Species: By means of natural selection or the preservation of favoured races in the struggle for life (No. 575.0162). Oxford University Press.
2. Dawkins, R. (1996). The blind watchmaker: Why the evidence of evolution reveals a universe without design. WW Norton & Company.
3. Futuyma, D.J. (2017). Evolution. Sinauer Associates Inc. USA.
4. Wilt, F. H., & Hake, S. (2004). Principles of developmental biology. W.W. Norton & Company, New York, USA.
5. Hall, B.K., & Hallgrimsson, B. (2017). Strasburger's Evolution. Jones and Bartlett Publishers, India.
6. Lewin, R. (2014). Human Evolution - An Illustrated Introduction. Wiley-Blackwell, USA.
7. Scott, F., & Gilbert, S.F. (2017). Developmental Biology. Sinauer Associates, Inc. USA.
8. Slack, J.M.W. (2015). Essential Developmental Biology, Wiley-Blackwell, USA.

# Foundation Course

**Course Code:** **MME.557**
**Course Title:** **Concepts of Bioinformatics**
**Total Hours:** **30**

| L | T | P | C |
|---|---|---|---|
| 2 | 0 | 0 | 2 |

**Course Learning Outcomes:**

On successful completion of the course the student will be able to:
CLO1: Analyze publicly available and experimental genomics data
CLO2: Perform in-silico prediction of protein structures and protein-protein interaction
CLO3: Perform in silico functional annotation of genetic findings

CLO4: Demonstrate the physical property of biomolecules in silico.

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| **Unit I** **6 Hours**<br>**Concept of biological databases:** Nucleotide Sequence Databases, GenBank, DDBJ, EMBL, Sequence Flat file and submission process, Protein sequence databases, UniProt in detail, Mapping databases, Genomic databases, Data mining. | **CLO1** |
| **UNIT II** **8 Hours**<br>**Inferring relationships:** Concept of global Vs. local sequence alignments, Dotplots, Scoring matrices, Pairwise sequence alignment, BLAST, Position-Specific scoring and PSI-BLAST, MegaBLAST, BL2SEQ, BLAT, FASTA Vs BLAST, Protein multiple sequence alignments, Basics of Sequence assembly and finishing. | **CLO2** |
| **UNIT III** **8 Hours**<br>**Sequence analysis:** Gene Prediction methods, Promoter analysis, RNA secondary structure thermodynamics, refining multiple sequence alignment based on RNA secondary structure predictions, SNP discovery methods and databases, Genotyping, International haplotype map project, 1000 genomes project. | **CLO3** |
| **Unit IV** **8 Hours**<br>**Analysis for protein sequences:** Predicting features of individual residues, Neural networks, Theory of Protein structure prediction, Protein structure databases, PDB in detail, Pathway and molecular interaction databases. | **CLO4** |

**Transactional Modes:** Lecture**;** Demonstration**;** Tutorial**;** Lecture cum demonstration; Problem solving; Case study; Self-learning.
**Internal assessment shall be through any of the following:** Surprise Tests, one sentence summary, case analysis, simulated problem solving, open book techniques, classroom assignments, homework assignments, term paper, Seminars, term papers, discussions, and presentations

**Suggested Readings:**

1. Baxevanis, A.D. and Ouellette, B.F.F. (2005). Bioinformatics: A Practical guide to the Analysis of Genes and Proteins. Wiley-Interscience, USA.
2. Hall, B.G. (2011). Phylogenetic Trees Made Easy: A How-To Manual. Sinauer Associates, Inc. USA.
3. Lesk, A.M. (2008). Introduction to Bioinformatics. Oxford University Press, UK.
4. Zvelebil, M. and Baum, J. (2007). Understanding Bioinformatics, Garland Science, New York, USA.
5. Ramsden, J. (2010). Bioinformatics: An Introduction (Computational Biology). Springer, India.
6. Ye, S.Q. (2008). Bioinformatics: A Practical approach. Chapman & Hall/CRC, UK.
7. Mount, D. (2012). Bioinformatics: Sequence and Genome Analysis. Cold Spring Harbor Laboratory Press.
8. Graur, D., Li, W. H. (2000). Fundamentals of Molecular Evolution. Sinauer Associates.
9. Tisdall, J. (2001). Beginning Perl for Bioinformatics. O'Really Publishers.

**Syllabus**
**M.Sc. in Human Genetics (Batch-2022)**

| | |
|---|---|
| **Course Code:** | **HGE.558** |
| **Course Title:** | **Innovation and Entrepreneurship** |
| **Total Hours:** | **15** |

| L | T | P | C |
|---|---|---|---|
| 1 | 0 | 0 | 1 |

**Course Learning Outcomes:**

On the completion of this course, the learners will:

CLO1: Understand the basic concepts of entrepreneur, entrepreneurship, and its importance.

CLO2: Aware of the issues, challenges, and opportunities in entrepreneurship.

CLO3: Develop capabilities of preparing proposals for starting small businesses.

CLO4: Know the availability of various institutional supports for making a new start-up.

| | |
|---|---|
| **Unit I** **4 Hours**<br>Entrepreneurial Structure; Nature, Characteristics, functions and its role in economic development Entrepreneurship- problems and prospects in India. Entrepreneurial Behavior and Skills. Entrepreneurial traits and skills; Types of entrepreneurial ventures; enterprise and society in Indian context; Importance of women entrepreneurship | **CLO1** |
| **Unit II** **4 Hours**<br>Role of industries/entrepreneur's associations and self-help groups; Funding opportunities for start-ups. Basic start-up problems; Preliminary contracts with the vendors, suppliers, bankers, principal customers; Contents of business plan/ project proposal. | **CLO2** |
| **Unit III** **4 Hours**<br>Promotion of a venture – Why to start a small business; How to start a small business; opportunity analysis, external environmental analysis, legal requirements for establishing a new unit, raising of funds, and establishing the venture<br>Blending University Research and Entrepreneurship culture, National Innovation and startup policy for students and faculty 2019 | **CLO3** |
| **Unit IV** **3 Hours**<br>Opportunities and challenges associated with establishing entrepreneurship in the field of Genetics and Molecular Medicine, Success stories | **CLO4** |

**Suggested Readings**:


1. Arora, Renu (2008). Entrepreneurship and Small Business, Dhanpat Rai & Sons Publications.
2. Chandra, Prasaaan (2018). Project Preparation, Appraisal, Implementation, Tata Mc-Graw Hills.
3. Desai, Vasant (2019). Management of a Small-Scale Industry, Himalaya Publishing House.
4. Jain, P. C. (2015). Handbook of New Entrepreneurs, Oxford University Press.
5. Srivastava, S. B. (2009). A Practical Guide to Industrial Entrepreneurs, Sultan Chand & Sons.
6. National Innovation and startup policy for students and faculty 2019, Government Policy document http://rmkcet.ac.in/RMK/NISP%20policy.pdf

**Syllabus**
**M.Sc. in Human Genetics (Batch-2022)**

| | |
|---|---|
| **Course Code:** | **HGE.600** |
| **Course Title:** | **Dissertation Part-I** |
| **Total Hours:** | **120** |

| L | T | P | C |
|---|---|---|---|
| 0 | 0 | 8 | 4 |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:

1. Apply genetic and genomics technique for research
2. Construct study design
3. Interpret result of a genetic experiment
4. Present oral and written scientific communication skills

The objective of dissertation part-I would be to ensure that the student learns the nuances of the scientific writing. Herein the student shall have to write his synopsis including an extensive review of literature with simultaneous identification of scientifically sound (and achievable) objectives backed by a comprehensive and detailed methodology. Student may start working in the respective laboratory.

Students can opt for dissertation work in industry, national institutes or Universities in the top 100 NIRF ranking. Group dissertation can be opted, with a group consisting of a maximum of four students. These students may work using a single approach or multidisciplinary approach. Research projects can be taken up in collaboration with industry or in a group from within the discipline or across the discipline

Synopsis will be evaluated as per the University policy.

**Evaluation criteria:**

| Examiner | Marks | Evaluation |
|---|---|---|
| Supervisor | 50 | Dissertation proposal and presentation |
| HoD and Senior-most faculty of the department | 50 | Dissertation proposal and presentation |

**Syllabus**
**M.Sc. in Human Genetics (Batch-2022)**

# Semester IV

| | |
|---|---|
| **Course Code:** | **HGE.601** |
| **Course Title:** | **Dissertation Part-II** |
| **Total Hours:** | **60** |

| L | T | P | C |
|---|---|---|---|
| 0 | 0 | 40 | 20 |

**Course Learning Outcomes:**
On successful completion of the course the student will be able to:

1. Apply genetic and genomics technique for research
2. Design research studies and perform research following scientific and ethical guidelines
3. Interpret results of genetic and molecular experiments
4. Present oral and written scientific literatures

During the course student will perform:

a. Synthesis of research hypothesis
b. Review of literature and identify research gap
c. Formulate methodology to achieve the objective of the research idea
d. Present articles and research ideas to fellow students and in other platforms
e. Perform researh and interpret the results
f. Write research reports and may publish research findings (if significant)

**Dissertation will be evaluated by the Department, as per the University policy.**
**Evaluation criteria**

| Examiner | Marks | Evaluation |
|---|---|---|
| Supervisor | 50 | Continuous assessment (regularity in work, mid-term evaluation) dissertation report, presentation, final viva-voce |
| External expert, HoD and senior-most faculty of the department | 50 | Dissertation report (30), presentation (10), final viva-voce (10) |

# Examination pattern for 2022-24 session

| **Core, Discipline Elective, Compulsory Foundation, Value Added and Interdisciplinary Courses** | | | **Discipline Enrichment Course** | | **Entrepreneurship Course** | |
|---|---|---|---|---|---|---|
| | **Marks** | **Evaluation** | **Marks** | **Evaluation** | **Marks** | **Evaluation** |
| Internal Assessment | 25 | Various | - | - | - | - |
| Mid-semester test (MST) | 25 | Subjective | 50 | Objective | 25 | Objective |
| End-semester test (EST) | 50 | Subjective (70%) Objective (30%) | 50 | Objective | 25 | Subjective |

| **Dissertation Proposal (Third Semester)** | | | **Dissertation (Fourth Semester)** | | |
|---|---|---|---|---|---|
| | **Marks** | **Evaluation** | | **Marks** | **Evaluation** |
| Supervisor | 50 | Dissertation proposal and presentation | Supervisor | 50 | Continuous assessment (regularity in work, mid-term evaluation) dissertation report, presentation, final viva-voce |
| HoD and senior-most faculty of the department | 50 | Dissertation proposal and presentation | External expert, HoD and senior-most faculty of the department | 50 | Dissertation report (30), presentation (10), final viva-voce (10) |

**Evaluation pattern similar to fourth semester dissertation will apply for internship where supervisor will award 50% marks and external co-supervisor, HoD and senior-most faculty will award 50% marks.**