INSTITUTE OF HUMAN GENETICS

SYLLABUS FOR POST GRADUATE PROGRAMME IN HUMAN GENETICS (2-YEAR M.Sc. COURSE) AS PER NATIONAL EDUCATION POLICY (NEP-2020)

SEMESTER-I

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

COURSE CODE	COURSE NAME	CREDITS
CR	EDIT FRAMEWORK FOR SEMESTE	R-I
	MAJOR CORE $[12 (T) + 6 (P)]$	
P2HGTC101	Cell and Molecular Biology	4
P2HGTC102	Physiology of Human Systems	4
P2HGTC103	Fundamentals of Genetics	4
P2HGPC104	Practical based on P2HGTC101	2
P2HGPC105	Practical based on P2HGTC102	2
P2HGPC106	Practical based on P2HGTC103	2
	18	
M	AJOR ELECTIVE (ANY ONE* 4T+2P	<u>')</u>
P2HGTE107	Biotechniques and Bioinstrumentation	4*
P2HGTE108	Microbial Genetics	4
P2HGPE109	Practical Based on P2HGTE107	2*
P2HGPE110	Practical Based on P2HGTE108	2
Total	6	
S	emester Credits Total	18+6= 24

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Semeste	r Course No.	Course Title	Credits	Assessment Scheme
I	P2HGTC101	Cell and Molecular Biology	Theory: 04 Practical: 02 Total: 06	Minor Test I: 10+10 Minor Test II: 10+10 Major test: 60 Total: 100

COURSE OUTCOMES:

CO1: Students will understand cell structure in prokaryotes and eukaryotes, including organelles and the cytoskeleton.

CO2: Students will learn cell cycle processes like mitosis, meiosis, and apoptosis, and how they are regulated.

CO3: Students will study DNA functions, including replication, transcription, translation, and related experiments.

CO4: Students will explore gene regulation and mutation types, along with DNA repair mechanisms.

CO5: Students will understand cell signaling and communication through various receptors and signal transduction pathways.

Unit I: Cell Structure and Function

12hours

- **1.1** Structure and Organization of Prokaryotic and Eukaryotic cell.
- **1.2** Plasma Membrane: Structure & transport across plasma membrane
- 1.3 Organelles and their functions (Nucleus, Mitochondria, ER, Golgi and Lysosomes)
- **1.4** Cytoskeleton and cell mobility: Microtubules, Microfilaments, Intermediate filaments

Unit II: Cell Cycle and its Regulation

- **2.1** Mitosis: Phases and Consequences.
- 2.2 Meiosis: Meiotic stages, synapsis, and Synaptonemal complex, Consequences.
- 2.3 Cell Cycle and its regulation: Cyclins & Cyclin dependent kinases, check points, Role of SMC, non-SMC, p53 & Rb proteins in regulation of cell cycle.
- **2.4** Programmed Cell Death (Apoptosis).

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Unit III: DNA Dynamics

12hours

- **3.1** DNA as a genetic material: Griffith transformation experiment, Avery MacLeod & McCarty Experiment & Hershey-Chase Experiment.
- **3.2** DNA replication: Prokaryotes & Eukaryotes.
- **3.3** DNA transcription and processing: Transcription in Prokaryotes & Eukaryotes, mRNA Capping, Polyadenylation, and mRNA splicing.
- **3.4** Protein synthesis and post translational modifications.

Unit IV: Gene Regulation & Genomic Maintenance

12hours

- **4.1** Gene regulation in prokaryotes and eukaryotes.
- **4.2** DNA replication: Prokaryotes & Eukaryotes.
- **4.3** Mutation and Mutagens: Physical, chemical & Biological mutagens; base substitutions, deletions & insertions, non-sense mutations, suppressor mutations, mis-sense mutations, silent mutations, frameshift mutations, germline and somatic mutations.
- **4.4** DNA repair mechanisms: Direct repair & Excision repair mechanisms (Base Excision repair, Nucleotide Excision repair, Mismatch Repair, Non- Homologous end Joining, SOS Repair)

Unit V: Cellular Interactions & Cell Signaling

- **5.1** Cell-Cell Interactions: Cell adhesions molecules, Cellular Junctions & Extracellular matrix.
- **5.2** Type of Cell Signaling: Autocrine, Paracrine, Endocrine, Juxtracrine.
- **5.3** Intracellular receptor and cell surface receptors.
- **5.4** Signal transduction pathways: G-Protein coupled receptor pathway, Receptor tyrosine kinase pathway, Janus Kinase Pathway, Insulin signaling and Mammalian Target of Rapamycin pathway and Secondary messengers.

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Books Recommended:

- 1. Alberts, B., Heald, R., Hopkin, K., Johnson, A., Raff, M., Roberts, K., & Walter, P. (2023). *Essential cell biology* (6th ed., International Student Edition). W. W. Norton & Company.
- **2.** Cooper, G. M., & Hausman, R. E. (2022). *The cell: A molecular approach* (9th ed.). Oxford University Press.
- 3. Alberts, B., Heald, R., Johnson, A., Morgan, D., Raff, M., Roberts, K., & Walter, P. (2020). *Molecular biology of the cell* (7th ed.). W. W. Norton & Company.
- **4.** Rastogi, S. C. (2023). *Cell biology* (5th ed.). New Age International Publishers.
- **5.** Krebs, J. E., Goldstein, E. S., & Kilpatrick, S. T. (2017). *Lewin's GENES XII* (12th ed.). Jones & Bartlett Learning.
- **6.** Brown, T. A. (2023). *Gene cloning and DNA analysis: An introduction* (8th ed.). Wiley-Blackwell.
- 7. Strachan, T., & Read, A. P. (2018). *Human molecular genetics* (5th ed.). Garland Science.
- 8. Snustad, D. P., & Simmons, M. J. (2015). Principles of genetics (7th ed.). Wiley.
- 9. Brooker, R. J. (2020). Genetics: Analysis and principles (7th ed.). McGraw-Hill Education.
- **10.** Cooper, G. M. (2019). *The cell: A molecular approach* (8th ed.). Oxford University Press.
- 11. Arumugam, N. (2019). Molecular biology and genetic engineering. Saras Publications.
- **12.** Cohn, R. D., Scherer, S. W., & Hamosh, A. (Eds.). (2024). *Thompson & Thompson genetics and genomics in medicine* (9th ed.). Elsevier.
- **13.** Karp, G., Iwasa, J., & Marshall, W. (2020). *Cell and molecular biology: Concepts and experiments* (9th ed.). Wiley.

Scheme of Examination: The students shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered in	Time allotted for	% Weightage
Subjective Test	the examination	the examination	(Marks)
Test I (after 30 days)	20%	1:30 hour	10+10 (20)
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)
Major test (after 90 days)	100%	3 hours	60
Total			100
Practicals/ Research			
Internal Examination	100%	2 hours	25
External examination	100%	2 hours	25
Total	1		50

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Note:

1. Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14 out of 40 will be eligible to re-appear in the Test I and Test II only once.

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

- Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.
- Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B.
 In major test there should not be a gap of more than two days in between two tests.

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PG SEMESTER-I

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

Semester	Course No.	Course Title	Credits	Assessment Scheme
I	P2HGTC102	Physiology of Human Systems	Theory: 04 Practical: 02 Total: 06	Minor Test I: 10+10 Minor Test II: 10+10 Major test: 60 Total:100

COURSE OUTCOMES:

CO1: Students will understand the structure and function of the human skeletal and muscular systems, including bone classification, muscle types, contraction mechanisms, and neuromuscular junctions.

CO2: Students will studythe human digestive and respiratory systems, covering anatomy, physiology, related hormones, and mechanisms of gas exchange and respiratory control.

CO3: Students will gain knowledge of cardiovascular and nervous systems, including heart function, neural anatomy, action potentials, and neurotransmission.

CO4: Students will explore the human excretory system, focusing on nephron structure, urine formation, and excretory physiology.

CO5: Students will learn the basics of endocrinology and reproduction, including major glands, hormone types, reproductive organs, and common endocrine disorders.

Unit-1 Human Musculo-Skeletal System

12hours

- **1.1** Bones and Cartilage: Classification, Histology, Ossification and Repair mechanisms.
- **1.2** Muscles Development, classification, and structure of different types of muscles.
- **1.3** Physiology of muscle contraction
- 1.4 Neuromuscular Junction

Unit-II Human Digestive & Respiratory System

- **2.1** Gross anatomy and physiology of Digestive System.
- **2.2** Hormones of the Digestive System.
- **2.3** Gross anatomy of the Respiratory system.
- **2.4** Physiology of Respiration: Exchange of oxygen and carbon dioxide, Transport of oxygen and Control of Respiration.

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Unit-III Human Cardiovascular & Nervous System

12hours

- **3.1** Anatomy & Physiology of the heart and peripheral vasculature.
- 3.2 Cardiac Cycle, Cardiac output and cardiac rhythms.
- **3.3**Structure of central and peripheral nervous System.
- 3.4 Action Potential and Neurotransmission

Unit-IV Human Excretory System

12hours

- **4.1** Gross anatomy of Excretory system.
- **4.2** Structure of Nephron.
- **4.3** Physiology of Excretion: Glomerular filtration, Tubular reabsorption and Tubular Secretion and urine production.
- **4.4** Homeostatic functions of the Human Excretory System.

Unit-V Human Endocrinology and Reproductive System

12hours

- **5.1** Overview of Endocrine Glands and types of hormones
- **5.2** Hypothalamus, Pituitary, Thyroid, Pancreas and adrenals.
- **5.3** Male and female reproductive system
- **5.4** Endocrine disorders.: Hypothyroidism, Hyperthroidism, Addison's disease, Cushing's syndrome, Congenital adrenal hyperplasia (CAH), Acromegaly & Gigantism.

Books Recommended:

- 1. Neal, J. M. (2016). How the endocrine system works (2nd ed.). Wiley-Blackwell.
- **2.** Tortora, G. J., & Derrickson, B. (2020). *Principles of anatomy and physiology* (16th ed.). Wiley.
- **3.** Melmed, S., Auchus, R. J., Goldfine, A. B., Rosen, C. J., & Kopp, P. A. (Eds.). (2024). Williams textbook of endocrinology (15th ed.). Elsevier.
- **4.** Chaurasia, B. D. (2023). *Handbook of general anatomy* (7th ed.). CBS Publishers & Distributors.

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- **5.** Sembulingam, K., & Sembulingam, P. (2019). *Essentials of medical physiology* (8th ed.). Jaypee Brothers Medical Publishers.
- **6.** Hall, J. E. (Ed.). (2020). *Guyton and Hall textbook of medical physiology* (14th ed.). Elsevier.
- 7. Norman, A. W., & Henry, H. L. (2022). *Hormones* (4th ed.). Academic Press.
- 8. Litwack, G. (2022). Hormones (4th ed.). Academic Press.

Scheme of Examination: The students shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered in	Time allotted for	% Weightage
Subjective Test	the examination	the examination	(Marks)
Test I (after 30 days)	20%	1:30 hour	10+10 (20)
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)
Major test (after 90 days)	100%	3 hours	60
Total			100
Practicals/ Research			
Internal Examination	100%	2 hours	25
External examination	100%	2 hours	25
Total	1		50

Note:

1. Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14 out of 40 will be eligible to re-appear in the Test I and Test II only once.

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

• Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.

UNIVERSITY OF JAMMU INSTITUTE OF HUMAN GENETICS SYLLABUS FOR POST GRADUATE PROGRAMME IN HUMAN GENETICS (2-YEAR M.Sc. COURSE) AS PER NATIONAL EDUCATION POLICY (NEP-2020)

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Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B.
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Semester	Course No.	Course Title	Credits	Assessment Scheme
I	P2HGTC103	Fundamentals of	Theory: 04	Minor Test I: 10+10
		Genetics	Practical: 02 Total: 06	Minor Test II: 10+10 Major test: 60
				Total:100

COURSE OUTCOMES:

CO1: Students will understand the structural organization of chromosomes in both prokaryotic and eukaryotic systems, including chromatin dynamics, nucleosome models, telomere architecture, and mitochondrial genome-related diseases.

CO2: Students will explore the principles of heredity and genetic variation, emphasizing Mendelian and non-Mendelian inheritance, gene interactions, and cytogenetic foundations of hereditary traits.

CO3: Students will examine chromosomal nomenclature and mechanisms of sex determination, including chromosomal landmarks, dosage compensation, and nonchromosomal determinants of sex.

CO4: Students will analyze genetic linkage and recombination processes, applying gene mapping techniques, LOD score analysis, and tetrad analysis to understand genetic distances and inheritance patterns.

CO5: Students will interpret patterns of genetic inheritance through pedigree analysis, while considering complex traits such as sex-linked inheritance, polygenic traits, and the influence of environmental and genetic variance on phenotypic expression.

UNIT I: Structure and Organization of Chromosomes

- **1.1** Structure of chromatin, heterochromatin, euchromatin.
- 1.2 Chromosome structure: Prokaryotes, Eukaryotes, Nucleosome model, Telomere structure, chromosome nomenclature & Landmarks of chromosomes
- **1.3** Specialized chromosomes: Lamp brush chromosomes and Polytene chromosomes.
- **1.4** Mitochondrial Genome and Associated diseases.

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Unit II: Hereditary and Modes of Inheritance

12hours

- **2.1**Chromosomal Theory of Heredity: Inheritance and Non disjunction as a proof to Chromosomal Theory.
- **2.2** Mendel's Laws of Inheritance: The Law of Dominance, the Law of Segregation & the Law of Independent Assortment.
- **2.3** Gene interactions and modifier genes.
- 2.4 Mitochondrial Inheritance.

Unit III: Genetic patterns of Inheritance

12hours

- **3.1** Mendelian patterns of Inheritance
- 3.2 Pedigree analysis: Symbols and Construction of a pedigree
- **3.3** Non-Mendelian modes of Inheritance: Exceptions of Mendelism: Codominance, Multipleallelism, Lethal alleles, Penetrance, Variable Expressivity, Pleiotropy etc. .
- **3.4**Quantitative Inheritance: Continuous & Discontinuous, Polygenic Inheritance, Genetic Variance & Heritability.

Unit IV: Sex Determination in Humans

12hours

- **4.1** Sex determination & differentiation
- **4.2** Inactivation of X chromosomes: Dosage compensation- Lyon Hypothesis
- **4.3** Sex-limited, sex linked and sex influenced traits
- **4.4** Non chromosomal basis of sex determination

Unit V: Genetic Linkage

- **5.1** Gene mapping: Two point and three-point test cross, LOD score analysis
- **5.2** Interference and Coincidence
- **5.3** Tetrad Analysis: Ordered tetrad and Unordered tetrad.
- **5.4** Recombination: Homologous and Non-Homologous recombination.

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Recommended Books

- **1.** Pyeritz, R. E., Korf, B. R., & Grody, W. W. (Eds.). (2024). *Emery and Rimoin's principles and practice of medical genetics and genomics* (7th ed.). Academic Press.
- **2.** Watson, J. D. (2024). *Molecular Biology of the Gene* (8th ed., multi-colour edition). Affiliated East West Press.
- **3.** Klug, W. S., Cummings, M. R., Spencer, C. A., Palladino, M. A., & Killian, D. (2025). *Concepts of Genetics* (13th ed.). Pearson.
- **4.** Lewis, R. (2024). *Human genetics: Concepts and applications* (14th ed.). McGraw-Hill Education.
- **5.** Cohn, R., Scherer, S., &Hamosh, A. (Eds.). (2023). *Thompson & Thompson genetics and genomics in medicine* (9th ed.). Elsevier.
- **6.** Pyeritz, R. E., Korf, B. R., & Grody, W. W. (Eds.). (2023). *Emery and Rimoin's principles and practice of medical genetics and genomics* (7th ed.). Academic Press.
- **7.** Brooker, R. J. (2020). *Genetics: Analysis and principles* (7th ed.). McGraw-Hill Education.
- **8.** Hartl, D. L., & Cochrane, B. J. (2017). *Genetics: Analysis of genes and genomes* (9th ed.). Jones & Bartlett Learning.
- **9.** Cummings, M. R. (2016). *Human heredity: Principles and issues* (11th ed.). Cengage Learning.
- **10.** Snustad, D. P., & Simmons, M. J. (2016). *Principles of genetics* (7th ed.). John Wiley & Sons.
- **11.** Griffiths, A. J. F., Wessler, S. R., Carroll, S. B., &Doebley, J. (2015). *An introduction to genetic analysis* (11th ed.). W. H. Freeman.
- 12. Kingston, H. M. (2002). ABC of clinical genetics (3rd ed.). BMJ Books.

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(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

Semester	Course No.	Course Title	Credits	Assessment Scheme
I	P2HGTE107	Biotechniques and Bioinstrumentation	Theory: 04 Practical: 02 Total: 06	Minor Test I: 10+10 Minor Test II: 10+10 Major test: 60
				Total: 100

COURSE OUTCOMES:

CO1: Students will comprehend core principles and applications of molecular separation and analysis techniques, including centrifugation, chromatography, electrophoresis, and spectrophotometry, for macromolecule isolation and quantification.

CO2: Students will understand and apply modern molecular biology techniques, such as PCR, gene cloning, genome editing (CRISPR-Cas9), and next-generation sequencing, for genetic analysis and manipulation.

CO3: Students will gain hands-on knowledge of bioinstrumentation tools like pH meters, microscopes, sterilization systems, and cell counters, essential for routine lab diagnostics and aseptic handling.

CO4: Students will explore advanced analytical technologies, including mass spectrometry, flow cytometry, electron/confocal microscopy, and microarrays, with emphasis on their roles in diagnostics, proteomics, and biomedical research.

CO5: Students will investigate structural biology and biosensing tools, such as NMR, X-ray crystallography, SPR, and lab-on-chip technologies, focusing on their diagnostic, therapeutic, and drug discovery applications.

Unit I: Biotechniques-I

12hours

1.1 Centrifugation Techniques: Principles of sedimentation (RCF, sedimentation coefficient), types such as differential and density gradient centrifugation, ultracentrifugation (preparative vs analytical), and applications in macromolecule isolation.

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- **1.2 Fundamentals of Chromatography:** Adsorption, partition, ion exchange, and size exclusion principles. Techniques include paper, thin-layer, column, and gel filtration chromatography with applications in protein and metabolite purification.
- **1.3 Electrophoresis Techniques:** Principles of electrophoresis, agarose gel and SDS-PAGE, DNA/RNA and protein separation, staining methods, and quantification using densitometry.
- **1.4 Spectrophotometry:** Beer-Lambert law, instrument components, nucleic acid/protein quantification, and enzyme kinetic assays.

Unit II: Biotechniques-II

12hours

- **2.1 Polymerase Chain Reaction (PCR):** Principles, types, primer design, amplification strategies, quantification, and troubleshooting.
- **2.2 Gene Cloning and Recombinant DNA Technology:** Restriction digestion, ligation, transformation, screening, and expression analysis in various systems.
- **2.3 Next-Generation Sequencing (NGS):** Overview of platforms (Illumina, Nanopore), library preparation, basic data analysis, and applications in genomics and transcriptomics.
- **2.4 CRISPR-Cas9 and Genome Editing:** Mechanism, guide RNA design, delivery methods, gene editing applications, and ethical issues.

Unit III: Bioinstrumentation-I

- **3.1 pH Meter and Electrochemical Instruments:** Principles, calibration, ion-selective electrodes, and enzyme kinetic applications.
- **3.2 Microscopy Fundamentals:** Light, phase contrast, dark field, and fluorescence microscopy with sample preparation and staining methods.
- **3.3 Autoclave, Laminar Flow, and Incubators:** Sterilization principles, biosafety protocols, and aseptic techniques.
- **3.4 Coulter Counter and Hemocytometer:** Principles, cell viability assays, and comparison of automated and manual counting.

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Unit IV: Bioinstrumentation –II

12hours

- **4.1 Mass Spectrometry:** Ionization (MALDI, ESI), mass analyzers, applications in proteomics and biomarker discovery.
- 4.2 Flow Cytometry and FACS: Principles, fluorochrome staining, data acquisition, and applications in cell sorting and immunophenotyping.
- **4.3 Confocal and Electron Microscopy:** Optical sectioning, 3D imaging, SEM and TEM sample prep and biological imaging.
- **4.4 Microarray Technology:** DNA/protein microarrays, probe hybridization, scanning, and expression profiling.

Unit V: Bioinstrumentation – III

12hours

- 5.1 NMR Spectroscopy: Principles, 1D/2D NMR techniques, sample handling, and applications in structure determination.
- 5.2 X-ray Crystallography: Crystallization, diffraction, instrumentation, and structure solving of proteins and nucleic acids.
- 5.3 Surface Plasmon Resonance (SPR) and Biosensors: Refractive index changes, kinetic analysis, and applications in drug discovery.
- **5.4 Lab-on-a-Chip and Microfluidics:** Microfabrication, fluid dynamics at microscale, integration with biosensors, and point-of-care diagnostics, ELISA-on-a-chip, Nucleic acid amplification tests (e.g., PCR, LAMP) on microfluidic platforms, Multiplexed biomarker detection.

Books Recommended:

- 1. Dubey, R. C., & Maheshwari, D. K. (2022). A textbook of biotechnology (5th rev. ed.). S. Chand Publishing.
- 2. Wilson, K., Walker, J. M., Hofmann, A., &Clokie, S. (2018). Wilson and Walker's principles and techniques of biochemistry and molecular biology (8th ed.). Cambridge University Press.

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SEMESTER-I

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

- **3.** Cappuccino, J. G., & Welsh, C. T. (2021). Microbiology: A laboratory manual (12th ed.). Pearson.
- **4.** Webster, J. G., &Nimunkar, A. J. (2020). Medical instrumentation: Application and design (5th ed.). Wiley.
- **5.** Hofmann, A., &Clokie, S. (Eds.). (2018). Wilson and Walker's principles and techniques of biochemistry and molecular biology (8th ed.). Cambridge University Press.
- **6.** Sambrook, J., & Green, M. R. (2012). Molecular cloning: A laboratory manual (4th ed.). Cold Spring Harbor Laboratory Press.
- **7.** Northrop, R. B. (2012). Analysis and application of analog electronic circuits to biomedical instrumentation (2nd ed.). CRC Press.
- 8. Miller, R. R., & Madsen, M. (2010). Microbiology: A laboratory manual. Pearson.
- **9.** de Hoffmann, E., &Stroobant, V. (2007). Mass spectrometry: Principles and applications (3rd ed.). Wiley.

Scheme of Examination: The students shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered in	Time allotted for	% Weightage
Subjective Test	the examination	the examination	(Marks)
Test I (after 30 days)	20%	1:30 hour	10+10 (20)
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)
Major test (after 90 days)	100%	3 hours	60
Total			100
Practical's/ Research			
Internal Examination	100%	2 hours	25
External examination	100%	2 hours	25
Total			50

Note:

1. Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who

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have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14

PG SEMESTER-I

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out of 40 will be eligible to re-appear in the Test I and Test IIonly once.

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

- Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.
- Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B.
 In major test there should not be a gap of more than two days in between two tests.

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SEMESTER-I

Semester	Course No.	Course Title	Credits	Assessment Scheme
I	P2HGTE108	Microbial Genetics	Theory: 04 Practical: 02 Total: 06	Minor Test I: 10+10 Minor Test II: 10+10 Major test: 60 Total: 100

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

COURSE OUTCOMES:

CO1: Students will gain the competence to demonstrate the fundamentals of microbiology, including its history, scope, and the structural and functional characteristics of bacteria, viruses, and fungi.

CO2: Students will be able to explain microbial genetic exchange mechanisms, such as transformation, conjugation, transduction, and phage genetics, and their applications in gene mapping.

CO3: Students will be able to describe plasmid and phage genetics, including the use of various vectors and molecular assays like EMSA and luciferase for gene regulation studies.

CO4: Students will explore major microbial pathogens, associated diseases, microbial markers, and the role of the human microbiome in health and disease.

CO5: Students will understand and apply microbial tools and techniques, including cloning, CRISPR, metagenomics, and reporter gene systems for genetic analysis and functional studies.

Unit I: Introduction to Microbiology

12hours

- **1.1** Introduction, history, and scope of Microbiology.
- **1.2** General characteristics and composition of Prokaryotes and Eukaryotes.
- **1.3** Morphology and ultra-structure of bacteria, Virus, and Fungus: size, shape, and arrangement
- **1.4** Toxins: Exotoxins and Endotoxins.

Unit II: Genetic Exchange Mechanisms

- **2.1** Transformation: natural competence, artificial transformation.
- **2.2** Conjugation: F plasmid, Hfr strains, mapping genes by interrupted mating.

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SEMESTER-I

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- **2.3** Transduction: generalized and specialized transduction.
- **2.4** Bacteriophage genetics: lytic and lysogenic cycles, genetic recombination in phages.

Unit III: Plasmid and Phage Genetics

12hours

- **3.1** Structure and function of plasmids.
- **3.2** Plasmid vectors: cloning, shuttle vectors, expression vectors, BACs, YACs.
- **3.3** Phage vectors: M13, lambda, cosmids.
- **3.4** EMSA and Luciferase assay.

Unit-IV Introduction to Pathogens

12hours

- **4.1** Bacterial Diseases (TB, Cholera, Pneumonia, Diarrhorea), Viral disease (AIDS, Hepatitis, COVID-19), Fungal disease (Candidiasis, Aspergillosis, blastomycosis)
- 4.2 Microbial markers
- **4.3** Microbiome, Gut micro biome and its function.
- **4.4** Microbiome in Human Diseases

Unit V Microbial Tools and Techniques

12hours

- **5.1** Molecular cloning techniques: restriction enzymes, ligation, transformation
- 5.2 Functional genomics: gene knockout, overexpression, CRISPR-Cas systems in microbes
- **5.3** Metagenomics studies: Introduction, applications, challenges and future directions.
- **5.4** Use of reporter genes (GFP, lacZ) and selectable markers

Books Recommended:

- 1. Mahon, C. R. (2024). Textbook of diagnostic microbiology (7th ed.). Elsevier.
- 2. Goering, R. V. (2024). Mims' medical microbiology and immunology (7th ed.). Elsevier.
- **3.** Murray, P. R., Rosenthal, K. S., & Pfaller, M. A. (2020). Medical microbiology (9th ed.). Elsevier.
- 4. Tortora, G. J., Funke, B. R., & Case, C. L. (2020). Microbiology (11th ed.). Pearson.
- **5.** Brooker, R. J. (2019). Genetics: Analysis and principles (6th ed.). McGraw-Hill Education.

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- 6. Strachan, T., & Read, A. P. (2018). Human molecular genetics (5th ed.). Garland Science.
- 7. Brock, T. D., Madigan, M. T., Martinko, J. M., & Parker, J. (2014). Brock biology of microorganisms (14th ed.). Pearson.
- Alberts, B., Johnson, A., Lewis, J., Raff, M., Roberts, K., & Walter, P. (2014). Molecular biology of the cell (6th ed.). Garland Science.
- 9. Brown, T. A. (2023). Genomes (5th ed.). CRC Press/Taylor & Francis.

Scheme of Examination: The students shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered in	Time allotted for	% Weightage
Subjective Test	the examination	the examination	(Marks)
Test I (after 30 days)	20%	1:30 hour	10+10 (20)
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)
Major test (after 90 days)	100%	3 hours	60
Total			100
Practicals/ Research			
Internal Examination	100%	2 hours	25
External examination	100%	2 hours	25
Total	1		50

Note:

Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14 out of 40 will be eligible to re-appear in the Test I and Test Honly once.

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.

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Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B.
 In major test there should not be a gap of more than two days in between two tests.

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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

COURSE CODE	COURSE NAME	CREDITS
C	REDIT FRAMEWORK FOR SEMESTER-	II
	MAJOR CORE $[12 (T) + 6 (P)]$	
P2HGTC201	Molecular Basis of Human Genetics	4
P2HGTC202	Human Embryology & Developmental	4
	Genetics	
P2HGTC203	Human Cytogenetics	2
P2HGTC204	Fundamentals of Biochemistry	2
P2HGPC205	Practical based on P2HGTC201	2
P2HGPC206	Practical based on P2HGTC202	2
P2HGPC207	Practical based on P2HGTC203/204	1+1=2
	18	
	MAJOR ELECTIVE (ANY ONE* 4T+2P)	
P2HGTE208	Research Design and Methodology	4*
P2HGTE209	Genetic Diagnostics	4
P2HGPE210	Practical Based on P2HGTE208	2*
P2HGPE211	Practical Based on P2HGTE209	2
Tot	6	
	18+6= 24	
EXIT OPTION		
P2HGVC251	Genetic Diagnostics and Genetic	4
	Counseling	

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(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

Semester	Course No.	Course Title	Credits	Assessment Scheme
II	P2HGTC201	Molecular Basis of HumanGenetics	Theory: 04 Practical: 02 Total: 06	Minor Test I: 10+10 Minor Test II: 10+10 Major test: 60 Total: 100

COURSE OUTCOMES:

CO1: Students will understand the structure and replication of nucleic acids, and explore key molecular techniques such as hybridization, recombinant DNA technology, and gene cloning with relevance to human diseases.

CO2: Students will develop the skill to analyse the molecular basis of genetic mutations and disorders, including nomenclature, loss/gain of function mutations, chromosomal abnormalities, and the role of epigenetics in human pathology.

CO3: Students will be able to evaluate gene identification strategies, such as positional and candidate gene approaches, mutation screening, and the implications of genome instability in disease development.

CO4: Students be prepared to explore large-scale genomic and proteomic projects, including the Human Genome Project and pangenome initiatives, and study transcriptomic and proteomic tools for understanding gene expression and regulation.

CO5: Students apply knowledge to examine modern diagnostics and therapeutic strategies, including gene therapy, genome editing (CRISPR), enzyme replacement therapy, and the use of model organisms in studying human genetic diseases.

Unit-INucleic Acid: Structure & Functions

- **1.1** Central Dogma of Life: DNA, RNA and Proteins
- **1.2** Methods and applications of molecular hybridization, synthesis and labelling of probes.
- 1.3 Gene Cloning- Types & Application, ethical and social issues in gene cloning.
- **1.4** Recombinant DNA Technology: Applications of RDT in Human Diseases.

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SEMESTER-II

Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

Unit-II: Genes and Molecular Pathology

12hours

- **2.1**Nomenclature for describing variation in human genome (types of variants/mutations) and Databases of mutations.
- **2.2**Human Genome Project, 1000 Genome Project, 100,000 Genome Project and Pan-genome Project.
- **2.3**From gene to disease and from disease to gene.
- **2.4** Epigenetics and its role in Human diseases.

UNIT -III: Gene and Disease

12hours

- **3.1**Discovering genes in diseases: Position independent and positional cloning.
- **3.2**Candidate gene approaches: Confirming a candidate gene through mutation screening.
- **3.3** Testing for a specified sequence change.
- **3.4** Instability of Human genome and Pathogenicity associated with repeated sequence.

UNIT-IV: Multi-Omics approach to human disease

12hours

- **4.1** Epigenomics and its significance in Human diseases.
- **4.2** Transcriptomics: Studying the Transcriptome by Microarray and Sequence Analysis.
- **4.3** Proteomics: Studying the Proteome and Protein Profiling (2D Electrophoresis & MALDI TOF).
- **4.4** Metagenomics and its applications in human diseases.

Unit- V: Therapeutics of Human Diseases

- **5.1** Animal models for the study of human genetic diseases (Drosophila, Yeast, Mouse, Neurospora).
- **5.2** Gene therapy: Concept and types of Gene therapy, Vectors used in gene therapy, (biological vectors retrovirus, adenovirus, herpes).
- **5.3** Gene Editing and genome editing treatments.
- **5.4** Stem cell therapy in single gene disorders.

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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

Recommended Books:

- 1. Watson, J. D. (2024). *Molecular biology of the gene* (8th ed.). Pearson.
- 2. Alberts, B. (2024). *Molecular biology of the cell* (7th ed.). Garland Science.
- **3.** Vaschetto, L. M. (Ed.). (2022). CRISPR-/Cas9 Based Genome Editing for Treating Genetic Disorders and Diseases (1st ed.). CRC Press/Taylor & Francis.
- **4.** Wilson, K., & Walker, J. (2018). *Biochemistry and molecular biology: Principles and techniques* (8th ed.). Cambridge University Press.
- **5.** Brown, T. A. (2017). *Genetics: A molecular approach* (4th ed.). CRC Press.
- **6.** Primrose, S. B. (2013). *Principles of gene manipulation and genomics* (7th ed.). Wiley-Blackwell.

Scheme of Examination: The students shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered in	Time allotted for	% Weightage
Subjective Test	the examination	the examination	(Marks)
Test I (after 30 days)	20%	1:30 hour	10+10 (20)
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)
Major test (after 90 days)	100%	3 hours	60
Total			100
Practical's/ Research			
Internal Examination	100%	2 hours	25
External examination	100%	2 hours	25
Total	1		50

Note:

1. Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14 out of 40 will be eligible to re-appear in the Test I and Test II only once.

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SEMESTER-II

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2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

- Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.
- Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B.
 In major test there should not be a gap of more than two days in between two tests.

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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

Semester	Course No.	Course Title	Credits	Assessment Scheme
II	P2HGTC202	Human Embryology & Developmental Genetics	Theory: 04 Practical: 02 Total: 06	Minor Test I: 10+10 Minor Test II: 10+10 Major test: 60 Total: 100

Course Objectives:

- **CO1**: Students will understand the basic molecular and cellular events in early human development.
- **CO2**: Students will learn the stages of embryonic and fetal development, including organ formation.
- **CO3**: Students will be able to explore how genes and cells guide tissue differentiation and body structure.
- **CO4**: Students will have the competence to examine how environmental factors and maternal health affect fetal development.
- **CO5**: Students will develop the skill to Identify and understand common congenital and reproductive developmental disorders.

Unit I: Cellular Foundations of Human Development

12hours

- **1.1** Fertilization and molecular events during fertilization.
- **1.2** Prenatal development of human embryo up to three germinal layers.
- **1.3** Differential gene activity and cell differentiation.
- **1.4** Gastrulation, cleavage patterns, fate map during gastrulation.

Unit II: Embryonic Development

12hours

- **2.1** Embryo Implantation.
- **2.2** Formation and types of placenta.
- **2.3** Notochord formation, Neurulation, development of nervous system.
- **2.4** Formation and Structure of somites.

Unit III: Organogenesis in Human Embryonic Development

12hours

3.1 Development of head and neck region.

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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

- **3.2** Formation of blood vessels, and development of cardiovascular system.
- **3.3** Development of limbs and skeletal system.
- **3.4** Development of gastrointestinal system, urogenital system and reproductive system.

Unit IV: Environment Factors in Development

12hours

- **4.1** Impact of paternal age and maternal health on fetal development.
- **4.2** Teratogens and developmental anomalies.
- **4.3**TORCH infections: effect on fetal development.
- **4.4** Epigenetics and environmental influences on the developing fetus.

Unit V: Common Congenital Malformations

12hours

- **5.1** Neural Tube Defects: Anencephaly, Encephalocele, Hydranencephaly, Spina Bifida
- **5.2** Cleft lip and palate
- **5.3**Congenital Heart Disease
- **5.4** Reproductive Disorders: Ambiguous genitalia, Gonadal dysgenesis, anomalies of genital duct, Hermaphroditism.

Books Recommended:

- **1.** Carlson, B. M. (2023). *Human embryology and developmental biology* (7th ed.). Elsevier.
- 2. Slack, J. M. W., & Dale, L. (2022). Essential Developmental Biology (4th ed.). Wiley-Blackwell.
- **3.** Devi, V. S. (Ed.). (2023). *Inderbir Singh's human embryology* (13th ed.). Jaypee Brothers Medical Publishers.
- **4.** Hoffman, B. L., Schorge, J. O., Bradshaw, K. D., Halvorson, L. M., Schaffer, J. I., & Corton, M. M. (2022). *Williamsgynecology* (4th ed.). McGraw-Hill Education.
- **5.** Sadler, T. W. (2021). *Langman's medical embryology* (14th ed.). Elsevier.
- **6.** Briggs, G. G., Freeman, R. K., & Towers, C. V. (2017). *Drugs in pregnancy and lactation: A reference guide to fetal and neonatal risk* (11th ed.). Wolters Kluwer.
- **7.** Barresi, M. J. F., & Gilbert, S. F. (2023). *Developmental Biology* (13th ed.). Sinauer Associates (Oxford University Press).

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SEMESTER-II

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- **8.** Moody, S. A. (Ed.). (2014). *Principles of developmental genetics* (2nd ed.). Elsevier.
- **9.** Shepard, T. H. (2010). *Catalog of teratogenic agents* (13th ed.). Johns Hopkins University Press.

Scheme of Examination: The students shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered in	Time allotted for	% Weightage	
Subjective Test	the examination	the examination	(Marks)	
Test I (after 30 days)	20%	1:30 hour	10+10 (20)	
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)	
Major test (after 90 days)	100%	3 hours	60	
Total			100	
Practical's/ Research				
Internal Examination	100%	2 hours	25	
External examination	sternal examination 100% 2 hours		25	
Total			50	

Note:

1. Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14 out of 40 will be eligible to re-appear in the Test I and Test II only once.

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

- Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.
- Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B.

In major test there should not be a gap of more than two days in between two tests.

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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

Semester	Course No.	Course Title	Credits	Assessment Scheme
П	P2HGTC203	Human Cytogenetics	Theory: 02 Practical: 01 Total: 03	Minor Test I: 5+5 Minor Test II: 5+5 Major test: 30 Total:50

COURSE OUTCOMES:

CO1: Students will develop the skill to identify and classify chromosomal abnormalities by recognizing structural changes (such as deletions, duplications, inversions, and translocations) and numerical changes (like aneuploidies) in human chromosomes.

CO2: Students will be able to understand the mechanisms underlying chromosomal abnormalities, including errors during cell division, particularly in meiosis, and their implications for human health.

CO3: Students gain the competence to explore the concept of genomic imprinting and its role in gene expression, highlighting how the parental origin of alleles can influence phenotypic outcomes.

Unit I: Chromosomal Anomalies

10hours

- **1.1** Introduction to human cytogenetics and related techniques.
- 1.2 Numerical Chromosomal Abnormalities: Down syndrome, Edward Syndrome & Patau Syndrome, Turner Syndrome & Klinefelter syndrome.
- 1.3 Structural Chromosomal abnormalities: Robertsonian and reciprocal translocations and its effect on human diseases.
- **1.4** Microdeletion syndromes: DiGeorge Syndrome, Cry-du-chat Syndrome, William-beuren Syndrome.

UnitII: Chromosome Breakage and Instability

- **2.1** Effects of chromosomal instability on human health.
- **2.2** Methods to study chromosomal breakage.
- 2.3 Chromosome Instability syndrome: Ataxia Telangiectasia, Fanconi Anemia, Bloom's Syndrome, Nijmegen breakage syndrome.
- **2.4** Chromosomal breakage and cancer susceptibility.

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UNIT-III: Cytogenetics and human disease

10hours

- **3.1** Cytogenetics of Leukemias
- **3.2** Cytogenetics of Solid Tumors.
- **3.3** Cytogenetics of recurrent pregnancy loss
- **3.4** Uniparental disomy and regions of homozygosity (ROH) as disease causing mechanisms.

Recommended Books:

- **1.** Cohn, R., Scherer, S., & Hamosh, A. (Eds.). (2024). *Thompson & Thompson genetics and genomics in medicine* (9th ed.). Elsevier
- **2.** Pyeritz, R. E., Korf, B. R., & Grody, W. W. (Eds.). (2019). Emery and Rimoin's principles and practice of medical genetics and genomics (7th ed.). Academic Press.
- 3. Korf, B. R., & Irons, M. B. (2014). Medical genetics (4th ed.). Wiley-Blackwell.
- 4. Mitelman, F., Mertens, F., & Johansson, B. (2015). Cancer Cytogenetics (4th ed.).
- 5. Gersen, S. L., & Keagle, M. B. (1999). The Principles of Clinical Cytogenetics.
- **6.** McKinlay Gardner, R. J., Sutherland, G. R., & Shaffer, L. G. (2011). *Chromosome Abnormalities and Genetic Counseling* (4th ed.). Oxford University Press.
- 7. Lewis, R. (2014). Human genetics: Concepts and applications (9th ed.). McGraw-Hill Education.
- 8. Gupta, R. C. (Ed.). (2022). Reproductive and developmental toxicology (3rd ed.). Elsevier
- 9. Ushakumari, R. (2020). Textbook on cytogenetics. New India Publishing Agency.
- **10.** Viville, S., & Sermon, K. D. (Eds.). (2023). *Textbook of human reproductive genetics*. Cambridge University Press.

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SEMESTER-II

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Scheme of Examination: The student shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered	Time allotted for the	% Weightage		
Subjective Test	in the examination	examination	(Marks)		
Test I (after 30 days)	20%	1 hour	05+05 (10)		
Test II (after 30 days)	21-40%	1 hour	05+05 (10)		
Major test (after 90 days)	100%	2.5 hours	30		
	50				
Practical/Research					
Internal Examination	100%	2hours	25		
External Examination	100%	2hours	25		
Total			50		

Note:

Major test will have two sections (A & B). Section A is compulsory comprising of 10 questions of 1 mark each and be spread over entire syllabus. Section B comprises of 4 questions from remaining 2 units and candidate must attempt one question from each unit of 10 marks each.

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II P2HGTC204 Fundamentals of Practical: 01 Minor Test Practical: 01 Total: 03 Minor Test Major test	st II: 10

Course objective:

CO1: Students will be able to explore inborn errors of metabolism, highlighting the genetic and biochemical basis, clinical features, and diagnostic approaches of disorders like galactosemia, phenylketonuria, and Tay-Sachs disease.

CO2: Students gain the competence to discuss the interdependence of metabolic pathways and how disruptions can lead to metabolic and systemic diseases, with emphasis on clinical correlations.

CO3: Equip students with diagnostic and interpretive skills to recognize metabolic disorders through symptomatology, lab findings, and appropriate management strategies.

Unit I: Carbohydrates Metabolism

10hours

- **1.1** Introduction to Biomolecules and their Classification.
- **1.2** Carbohydrates: Structure, Types & functions.
- **1.3** Carbohydrates metabolism: Glycolysis and Gluconeogenesis.
- **1.4** Metabolic disorders of Carbohydrates (Pathophysiology, diagnosis, and Management): Lactose & fructose Intolerance, Galactosemia, Diabetes Mellitus, Glucose-6-phosphate dehydrogenase deficiency.

Unit II: Protein and Nucleic acid metabolism

- **2.1** Proteins, amino acids & Nucleic acids: Structure, types & functions.
- **2.2** Protein Metabolism: Deamination, transamination & Urea cycle.
- **2.3** Nucleic Acids metabolism: De Novo Synthesis and Salvage pathways.
- **2.4** Metabolic disorders of proteins, amino acids and of Purines and Pyrimidines (Pathophysiology, diagnosis, and Management): Phenylketonuria, Alkaptonuria, Albinism, Maple Syrup urine disease, Hyperuricemia & Lesch-Nyhan Syndrome & Adenosine deaminase deficiency.

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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

Unit III: Lipid metabolism

10hours

- **3.1** Lipids: Structure, Types & Functions.
- 3.2 Fatty Acid Synthesis and Shuttle Systems
- **3.3** Lipid Catabolism and Oxidation Pathways
- **3.4** Lipid Metabolism Disorders (Pathophysiology, Diagnosis, and Management): Hypercholesterolemia, Niemann-Pick Disease, Tay-Sachs disease, Gaucher disease.

Books Recommended:

- 1. Stryer, L., Berg, J. M., Tymoczko, J. L., & Gatto, G. J. Jr. (2023). Biochemistry (10th ed.). W. H. Freeman.
- 2. Berg, J. M., Gatto, G. J. Jr., Hines, J. K., Tymoczko, J. L., &Stryer, L. (2023). *Biochemistry* (10th ed.). W. H. Freeman.
- 3. Nelson, D. L., & Cox, M. M. (2021). Lehninger principles of biochemistry (9th ed.). W. H. Freeman.
- **4.** Voet, D., Voet, J. G., & Pratt, C. W. (2018). Voet's principles of biochemistry (5th ed., global ed.). Wiley.
- 5. Wilcox, W. D., & Sly, W. S. (2010). Inborn errors of metabolism (4th ed.). Springer.
- 6. Devlin, T. M. (2018). Textbook of biochemistry with clinical correlations (8th ed.). Wiley-Liss
- 7. Rodwell, V. W., Bender, D. A., Botham, K. M., Kennelly, P. J., & Weil, P. A. (2022). Harper's illustrated biochemistry (32nd ed.). McGraw-Hill Education.
- **8.** Salway, J. G. (2017). Metabolism at a glance (4th ed.). Wiley-Blackwell.

Scheme of Examination: The student shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered	Time allotted for the	% Weightage	
Subjective Test	in the examination	examination	(Marks)	
Test I (after 30 days)	20%	1 hour	05+05 (10)	
Test II (after 30 days)	21-40%	1 hour	05+05 (10)	
Major test (after 90 days)	100%	2.5 hours	30	
	50			
Practical/Research				
Internal Examination	100%	2hours	25	
External Examination	100%	2hours	25	
Total			50	

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SEMESTER-III

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Note:

Major test will have two sections (A & B). Section A is compulsory comprising of 10 questions of 1 mark each and be spread over entire syllabus. Section B comprises of 4 questions from remaining 2 units and candidate must attempt one question from each unit of 10 marks each.

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SEMESTER-II

Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027

Semester	Course No.	Course Title	Credits	Assessment Scheme
II	P2HGTE208	Research Design and Methodology	Theory: 04 Practical: 02 Total: 06	Minor Test I: 10+10 Minor Test II: 10+10 Major test: 60
				Total: 100

COURSE OUTCOMES:

CO1: Students will be able to Introduce the fundamentals of scientific research, including formulation of research problems, hypotheses, and objectives.

CO2: Students will develop skills in scientific writing and publishing, including manuscript preparation, citation, editing, and ethical considerations like plagiarism and conflict of interest.

CO3: Familiarize students with various research study designs such as cohort, case-control, cross-sectional, and clinical trials, along with their applications and limitations.

CO4: Students acquire knowledge and skills for designing of effective research tools, including questionnaires, consent forms, and appropriate sampling strategies with power and sample size calculations.

CO5: Promote safe laboratory practices, emphasizing proper specimen handling, biosafety protocols, and management of hazardous materials in biomedical research.

Unit I: Basics of Research and Scientific Writing

12hours

- 1.1 Research Problem, Objectives & Hypothesis,
- **1.2** Uses of Sources of Information such as journals, books, index medicus, Excerpta Medica, Biological abstracts, Science Citation Index.
- **1.3** Preparing a manuscript for publication.
- **1.4** Editing and Galley proof correction of manuscripts.

Unit II: Research Essentials

- **2.1** Publication Ethics, Conflicts of Interest, Plagiarism.
- **2.2** Protocol Content for research project.
- **2.3** Preparation and Delivering of Scientific talk/ putting up poster displays.

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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

2.4 References and their citation.

Unit III: Study Designs in Research

12hours

- **3.1** Cohort & Longitudinal studies with merits and demerits.
- 3.2 Case Control and Nested Case Control study with merits and demerits, metanalysis.
- **3.3** Cross-sectional studies with merits and demerits
- **3.4** Familial/Linkage studies, Clinical trials, and their utility in Research

Unit IV: Questionnaires and Sampling Methods

12hours

- **4.1** Designing of Questionnaire and Consent form
- **4.2** Sampling concept, advantage, limitation, and precautions of sampling.
- **4.3** Methods of sampling: Random &Non random, References and their citation.
- **4.4** Sample size determination: power of study and its calculation in research.

Unit V: Specimen Handling & Lab Safety

12hours

- **5.1** Collection, Transportation, and storage of various biological materials
- **5.2** Hazardous and radiolabeled chemicals and their handling.
- **5.3** Decontamination and disposal methods.
- **5.4** Biosafety Levels, Safety checklist for biomedical labs.

Books Recommended:

- 1. Kothari, C. R., & Garg, G. (2023). Research methodology: Methods and techniques (5th ed.). New Age International Publishers.
- **2.** Creswell, J. W., & Creswell, J. (2022). *Research Design: Qualitative, Quantitative, and Mixed Methods Approaches* (6th ed.). SAGE Publications.
- **3.** Gurumani, N. (2021). Research methodology for biological sciences. MJP Publishers.
- **4.** Indrayan, A., & Malhotra, R. K. (2017). Medical biostatistics (4th ed.). Chapman & Hall/CRC Press.
- **5.** Ford, E. D. (2000). Scientific methods in ecological research. Cambridge University Press.
- **6.** Dear, P. H. (Ed.). (1997). Genome mapping. IRL Press / Oxford University Press.

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SEMESTER-II

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- 7. Hawkins, C., & Sorgi, M. (1985). Research How to plan, speak and write about it. Springer-Verlag.
- **8.** Singh, R., Singh, A. K., Mishra, S., Kumari, S., & Yadav, M. (2024). *Research methodology in life sciences* (Vol. 1). Natals Publication.

Scheme of Examination: The students shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered in	Time allotted for	% Weightage				
Subjective Test	the examination	the examination	(Marks)				
Test I (after 30 days)	20%	1:30 hour	10+10 (20)				
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)				
Major test (after 90 days)	100%	3 hours	60				
Total			100				
Practical's/ Research	Practical's/ Research						
Internal Examination	100%	2 hours	25				
External examination	100%	2 hours	25				
Total	1		50				

Note:

1. Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14 out of 40 will be eligible to re-appear in the Test I and Test II only once.

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

- Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.
- Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B.

In major test there should not be a gap of more than two days in between two tests.

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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

Semester	Course No.	Course Title	Credits	Assessment Scheme
II	P2HGTE209	Genetic Diagnostics	Theory: 04 Practical: 02 Total: 06	Minor Test I: 10+!0 Minor Test II: 10+10 Major test: 60 Total: 100

COURSE OUTCOMES:

- **CO1**: Students will understand the principles and applications of molecular diagnostic techniques, including PCR, DNA sequencing, SNP analysis, and screening methods for genetic and disease diagnosis.
- **CO2**: Students will explore cytogenetic techniques such as karyotyping, FISH, MLPA, CGH, and GISH, and their role in detecting chromosomal abnormalities.
- CO3: Students will learn fundamental laboratory methods including centrifugation, electrophoresis, spectrophotometry, microscopy, blotting, and chromatography, and their applications in molecular diagnostics.
- **CO4**: Students will examine prenatal diagnostic methods, both invasive and non-invasive, and the role of NGS and point-of-care devices in early disease detection.
- CO5: Students will gain competence to Stay informed about emerging technologies in molecular diagnostics, including the latest advancements in sequencing platforms and diagnostic tools.

UNIT I: Introduction to Genetic Diagnostics

12hours

- 1.1 History, significance, and scope of genetic diagnostics
- **1.2** Rise of Genomic Diagnostic Industry in Indian and Global scenario.
- **1.3** Chromosomal and Molecular techniques, DNA and RNA sequencing, In situ hybridization, Microarray analysis, mass spectroscopy
- **1.4**Advances in Biochemistry, Histopathology, Flow cytometry and Imaging based diagnosis.

UNIT II: Basic Techniques and applications in Laboratory Diagnostics 12hours

2.1 Centrifugation and Electrophoresis: Basic principle, Types and Applications.

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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

- **2.2** Spectrophotometer: Principle, Working and Applications.
- **2.3** Microscopy: Types, Principle, Working, Application.
- **2.4** Blotting techniques (Northern, Southern and Western Blotting and Dot Blot assay) and Chromatography techniques (Paper, Liquid, Gas, and Thin Layer Chromatography).

UNIT III: Classical Cytogenetics and Molecular Cytogenetic Techniques 12hours

- **3.1** Overview of Cytogenetic Techniques.
- **3.2** Conventional Karyotyping: Lymphocyte culture, harvesting and banding, Special Chromosome banding techniques.
- **3.3** Molecular Cytogenetic Techniques: Fluorescence In Situ Hybridization (FISH), Spectral Karyotyping and Genomic in Situ Hybridization(GISH), Optical Genome Mapping (OGM).
- **3.4** Comparative Genomic Hybridization (CGH) and Chromosomal Microarray.

UNIT IV: Molecular Diagnostic Techniques

12hours

- **4.1** PCR: Types and applications (Real time-PCR, Multiplex PCR, reverse transcriptase-PCR, Nested PCR, RFLP, AFLP and Isothermal PCR).
- **4.2** DNA Sequencing: Maxam-Gilbert (Chemical) and Sanger sequencing method.
- **4.3** Massive parallel sequencing (NGS and pyrosequencing)/SNPs (rsIDS) and SNP databases.
- **4.4** Multiplex Ligation probe dependent Microarray (MLPA-/Digital MLPA.

UNITV: Prenatal and Diagnostic Techniques

- **5.2** Prenatal diagnostics: Screening for genetic disorders.
- **5.2** Non-invasive techniques- Ultrasonography, maternal fetal serum, and fetal cells in the maternal blood, NIPT (Cell free fetal DNA).
- **5.3** Invasive techniques: Chorionic Villi Sampling, Amniocentesis, Fetoscopy.
- **5.4** Carrier screening and population screening for genetic disorders.

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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec 2025, Dec 2026 & Dec 2027)

Books Recommended:

- 1. Alberts, B., Heald, R., Johnson, J., Morgan, D., Raff, M., Roberts, K., & Walter, P. (Eds.). (2022). Molecular biology of the cell (7th ed.). W. W. Norton & Company.
- **2.** Cooper, G. M., & Adams, K. (2022). The cell: A molecular approach (9th ed.). Sinauer Associates (Oxford University Press).
- **3.** Pyeritz, R. E., Korf, B. R., & Grody, W. W. (Eds.). (2019). Emery and Rimoin's principles and practice of medical genetics and genomics (7th ed.). Academic Press.
- **4.** Brown, T. A. (2023). Genomes (5th ed.). CRC Press.
- 5. Krebs, J. E., Goldstein, E. S., & Kilpatrick, S. T. (Eds.). (2017). Lewin's genes (12th ed.). Jones & Bartlett Learning.
- **6.** Malhotra, N., Kumar, P., Malhotra, J., Malhotra Bora, N., & Mittal, P. (2019). *Jeffcoate's Principles of Gynaecology* (9th ed.). Jaypee Brothers Medical Publishers.

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Subjective Test	the examination	the examination	(Marks)
Test I (after 30 days)	20%	1:30 hour	10+10 (20)
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)
Major test (after 90 days)	100%	3 hours	60
Total			100
Practical's/ Research			
Internal Examination	100%	2 hours	25
External examination	100%	2 hours	25
Total	1		50

Note:

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(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

- Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.
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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

Semester	Course No.	Course Title	Credits
II	P2HGVC251	Genetic Diagnostics and Genetic Counseling	Theory: 04 Total: 04

COURSE OUTCOMES:

- **CO1**: Students will understand core molecular and cytogenetic techniques including DNA isolation, lymphocyte culture, karyotyping, and diagnosis of chromosomal abnormalities.
- **CO2**: Students will be able to analyze genetic variants and interpret sequencing data using advanced technologies such as MLPA, NGS, and Optical Genome Mapping.
- **CO3**: Students will develop skill to Utilize genomic databases and bioinformatics tools for variant classification, annotation, and interpretation in clinical genetics.
- CO3: Students will be able to demonstrate knowledge and application of genetic counseling principles, including ethical considerations, pedigree analysis, and patient interaction.
- CO5: Students will gain knowledge to evaluate ethical, legal, and social implications (ELSI) of genetic testing and counseling, focusing on regulations, consent, and psychosocial aspects.

UNIT I: Cytogenetics and Molecular Techniques

12hours

- **1.1** Basic Techniques in Molecular Biology: DNA isolation & Quantification of DNA.
- **1.2** Lymphocyte culture: Harvesting and Scanning.
- **1.3** Karyotyping and identification of common chromosomal syndromes; Down syndrome, Edward syndrome, Patau syndrome, Turner syndrome, & Klinefelter syndrome.
- **1.4** Diagnosis of microdeletion-microduplication syndromes by FISH/chromosomal microarray.

UNIT II: Molecular Genetics (Sequencing and Variant classification)12hours

2.1 Types of genetic variants: SNV's, InDels, CNVs and structural variants and use of direct and indirect DNA testing methods.

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SEMESTER-II

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

- 2.2 High throughput diagnostics tests: Multiplex Ligation-dependent Probe Amplification (MLPA), Next Generation Sequencing (NGS), Optical Genome Mapping (OGM).
- **2.3** Variant Classification: ACMG- ACP guidelines/ ACGS Best practice guidelines.
- **2.4** Identification of Syndromes using AI based tools: Gestalt Matcher/ Face2Gene.

UNIT III: Genomic Databases and Bioinformatics Tools

12hours

- **3.1** Introduction to Genetic Variant Databases: Purpose and significance.
- 3.2 Genetic databases: OMIM, HapMap Project, ClinVar, Human Gene Mutation Database (HGMD), dbSNP, 1000 Genomes Project, Exome Aggregation Consortium (ExAC) / gnomAD, LOVD (Leiden Open Variation Database), DECIPHER.
- **3.3**Variant analysis tools: GATK/VARSOME/ANNOVAR
- **3.4** Generative AI in Clinical Genetics- Advantages and Limitations.

UNIT IV: Genetic Counseling

12hours

- **4.1** Genetic Counseling: Scope, process, and prenatal/pre-conceptional genetic counselling.
- **4.2** Code of Ethics in Genetic Counseling: Autonomy, beneficence, justice, informed consent.
- **4.3** Pedigree analysis, inheritance patterns, and study of traits (tongue rolling, ear lobe, widow's peak, eye color).
- **4.4** Case studies and clinical interactions: OPD visits, Phenotype identification, Psychosocial aspects.

UNIT V: ELSI (Ethical- Legal-Social Issues) related to Genetic testing and **Counseling**

- 5.1 Risks and benefits of Genetic testing, Informed decision making and Psychosocial implications of Genetic testing results for the family.
- **5.2** Regulation of prenatal diagnostic techniques: the PNDT and PC-PNDT Acts [Prenatal Diagnostic techniques (Regulation and prevention of misuse) Act, 1994- preconception

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SEMESTER-III

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

Prenatal Diagnostic techniques (Prohibition of sex selection) Act]

- **5.3**Registration and Regulation of Genetic Counseling Centres, Genetic Laboratories and genetic Clinics, appropriate authority & Advisory committees
- **5.4**Consent for testing asymptomatic minor, predictive testing for cancers in asymptomatic individuals.

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SEMESTER-III

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COURSE CODE	COURSE NAME	CREDITS					
CREDIT FRAMEWORK FOR SEMESTER-III							
	Major Core [14 (T) + 6 (P)]						
P2HGTC301	Medical Genetics & Genomics	4					
P2HGTC302	Population & Evolutionary Genetics	4					
P2HGTC303	Basics of Bioinformatics & Biostatistics	2					
P2HGTC304	Fundamentals of Clinical Genetics	4					
P2HGPC305	Practical based on P2HGTC301	2					
P2HGPC306	Practical based on P2HGTC302	2					
P2HGPC307	Practical based on P2HGTC303	2					
	Total Credits	20					
	MAJOR ELECTIVE (ANY ONE*) (2T+2P)						
P2HGTE308	Stem Cell Technology	2*					
P2HGTE309	Cancer Biology	2					
P2HGPE310	Practical Based on P2HGTE307	2*					
P2HGPE311	Practical Based on P2HGTE308	2					
	4						
P2HGMO351	MOOC/SWAYAM COURSES	4					
	Semester Credits Total	20+4+4= 28					

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SEMESTER-III

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

Semester	Course No.	Course Ti	tle		Credits	Assessment Scheme
III	P2HGTC301	Medical Genomics	Genetics	&	Theory: 04 Practical: 02 Total: 06	Minor Test I: 10+10 Minor Test II: 10+10 Major test: 60
						Total: 100

COURSE OUTCOMES:

CO1: Students will be able to Illustrate an in-depth understanding of the spectrum and inheritance patterns of genetic diseases, including autosomal, sex-linked, mitochondrial, and multifactorial disorders.

CO2: Apply modern genomic technologies such as NGS, WGS, and WES for the detection, screening, and management of inherited and somatic genetic diseases.

CO3: Interpret and analyze principles of personalized medicine, including pharmacogenomics, nutrigenomics, and preventive genomics, in the context of clinical applications.

CO4: Evaluate the molecular basis of cancer by identifying oncogenes, tumor suppressor genes, and genomic alterations, and assess the role of oncogenomics in diagnostics and ethical clinical practices.

CO5: Examine emerging technologies in medical genetics, including synthetic biology, transcriptomics, metabolomics, and reproductive technologies, along with their ethical and intellectual property considerations.

UNIT I: Inheritance of Genetic Diseases/Disorders

- **1.1** Spectrum of genetic diseases (single gene, chromosomal, multifactorial, mitochondrial, somatic cell genetic diseases) and their patterns of inheritance.
- **1.2** Inheritance pattern of autosomal single gene disorders: Sickle cell anemia, Huntington disease, Cystic Fibrosis, Neurofibromatosis. Marfan syndrome
- **1.3** Inheritance pattern of sex-linked single gene disorders: Hemophilia, DMD & Fragile-X syndrome.

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SEMESTER-III

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

1.4 Inheritance pattern of mitochondrial diseases: Mitochondrial Myopathy, Diabetes mellitus and deafness (DAD), Leber's hereditary optic neuropathy (LHON), Myoclonic epilepsy with ragged red fibers (MERRF syndrome).

UNIT II: Detection and Management of Genetic Diseases

12hours

- **2.1** Detection of inherited genetic diseases by NGS: Whole genome sequencing (WGS), and whole exome sequencing (WES).
- **2.2** Reproductive Genomics: Screening and prenatal diagnosis of genetic disorders.
- **2.3** Cancer Theranostics: Diagnostic biomarkers and precision therapeutic agents.
- **2.4** Management & Treatment of genetic diseases.

UNIT III: Personalized medicine in clinical practice

12hours

- 3.1 Introduction to pharmacogenomics, Variability in drug metabolism and side effects and methods of analysis in pharmacogenomics.
- **3.2** Personalizing the right dose: antipsychotics, antidepressants, antiepileptics, antidiabetics, cardiovascular drugs, NSAIDs, analgesic drugs and immunosuppresants, Nanotechnology in medicine.
- 3.3 Nutrigenomics: the relationship between human genome, human nutrition, and health.
- **3.4** Preventive genomics: Predictive risks, scores for complex disorders.

UNIT IV: Oncogenomics

12hours

- 4.1 Oncogenes (e.g., MYC, KRAS, BCR-ABL) and tumor suppressor genes (e.g., TP53, RB1, BRCA1/2).
- **4.2** Hallmarks of cancer and genomic instability.
- **4.3** Major cancer genomics initiatives: TCGA, ICGC, COSMIC.
- 4.4 Use of genomic biomarkers for diagnosis and prognosis, ethical issues in cancer genomics.

UNIT V: Innovative Technologies in Medical Genetics

12hours

5.1 Historical developments in Medical Genetics.

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SEMESTER-III

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

- **5.2** Assisted Reproductive Technologies, Somatic Cell Nuclear transplant & its applications.
- **5.3** Synthetic biology and its applications, Introduction to IPR.
- **5.4** Applications of Transcriptomics and Metabolomics in Medical Genetics.

Books Recommended:

- **1.** Descartes, R. J., & Cheng, T. L. (2020). Thompson & Thompson genetics in medicine (9th ed.). Elsevier.
- **2.** Wang, W., et al. (2022). Pharmacogenomics: Challenges and opportunities in therapeutic strategies. Academic Press.
- **3.** Khoury, M. J., et al. (Eds.). (2021). Genomic and precision medicine: Ethics, challenges, and opportunities. Elsevier.
- 4. Baldwin, G., & Endy, D. (2023). Synthetic biology: A primer (2nd ed.). CRC Press.
- **5.** Thakur, A., Thakur, P., & Suhag, D. (2025). Advancements in nanobiology characterization techniques and cutting-edge applications (1st ed.). Academic Press.
- **6.** Cao, G., & Wang, W. (2011). Nanostructures and nanomaterials: Synthesis, properties (2nd ed.). Imperial College Press.
- **7.** Saharan, V., & Pal, A. (2016). Chitosan-based nanomaterials in plant growth and protection. Springer Briefs in Nanoscience and Nanotechnology.

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Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)			
Major test (after 90 days)	100%	3 hours	60			
Total		100				
Practicals/ Research	Practicals/ Research					
Internal Examination	100%	2 hours	25			
External examination	100%	2 hours	25			
Total	1	1	50			

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SEMESTER-III

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

Note:

1. Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14 out of 40 will be eligible to re-appear in the Test I and Test II only once.

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

- Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.
- Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B. In major test there should not be a gap of more than two days in between two tests.

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SEMESTER-III

Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028

Semester Course No. Cour	Title Credits Assessment Scheme
III P2HGTC302 Popul Evolu	onary Genetics Theory: 04 Practical: 02 Total: 06 Minor Test I: 10+10 Major test: 60 Total:100

COURSE OUTCOMES:

- **CO1**: Students will understand the fundamental principles of biological evolution, including natural selection, speciation, and human evolutionary pathways.
- **CO2**: Students will be able to learn the concepts and tools of phylogenetics, with emphasis on molecular phylogeny, comparative genomics, and the molecular clock.
- **CO3**: Students will develop the skill to explain the genetic structure of populations, using models like Hardy-Weinberg Equilibrium and various genetic association study designs.
- **CO4**: Students will explore the roles of mutation, selection, and genetic variation in shaping populations, including estimation techniques and adaptive mechanisms.
- CO5: Students will analyze the effects of inbreeding, genetic drift, and migration, including calculations of inbreeding coefficients and implications for genetic health and diversity.

Unit I: Evolutionary Biology

12hours

- **1.1** Biological Evolution and Theories of Evolution
- **1.2** Natural selection: Evidences and modes of Natural selection, sexual selection.
- **1.3** Speciation: types of speciation, evolutionary processes causing speciation
- **1.4** Evolution of Modern Humans.

Unit II: Phylogenetics

- **2.1** Concepts and applications of Phylogenetics
- **2.2** Molecular phylogeny: Phylogenetic tree construction
- **2.3** Molecular clock
- **2.4** Comparative Genomics.

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Unit III: Population Genetics

12hours

- **3.1** Genetic constitution of a population, calculation of allele and genotype frequencies.
- **3.2** Types of genetic association studies: Case control, Cross-sectional, GWAS, quantitative trait association studies.
- **3.3** Hardy-Weinberg Equilibrium Principle: Assumptions of HWE, Testing of HWE in the population, HWE for two and multiple alleles of an autosomal gene.
- **3.4** Impact of recurrent and non-recurrent mutations in HWE population.

Unit IV: Genetic Dynamics: Mutation, Selection, and Variation

12hours

- **4.1** Mutation pressure and estimates of mutation rates.
- **4.2** Selection coefficient and fitness, Selection against recessive/ dominant/partial dominant/overdominant genes, heterozygote advantage
- **4.3** Phenotypic variation and spatial variation.
- **4.4** Adaptive landscape, gene pool & genetic fitness, Bayes theorem.

Unit V: Inbreeding, Genetic Drift, and Migration: Impacts on Populations 12hours

- **5.1** Consanguinity and inbreeding.
- **5.2** Inbreeding coefficient of a population and individual through path analysis.
- **5.3** Biological consequences of inbreeding, genetic load and its measurements.
- **5.4** Effect of migration and genetic drift on gene frequencies.

Recommended Books:

- **1.** Cohn, R. D., Scherer, S. W., & Hamosh, A. (Eds.). (2023). *Thompson & Thompson genetics and genomics in medicine* (9th ed.). Elsevier.
- **2.** Rastogi, V. B. (2023). *Organic evolution: Evolutionary biology* (15th ed.). Medtech Science Press.
- 3. Hamilton, M. B. (2021). *Population genetics* (2nd ed.). Wiley-Blackwell.
- **4.** Knopik, V. S., Neiderhiser, J. M., DeFries, J. C., & Plomin, R. (2016). *Behavioral genetics* (7th ed.). Worth Publishers.
- **5.** Hedrick, P. W. (2011). *Genetics of population* (4th ed.). Jones and Bartlett Publishers.

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- **6.** Hartl, D. L., & Clark, A. G. (2007). *Principles of population genetics* (4th ed.). Sinauer Associates.
- 7. Relethford, J. H. (2012). *Human population genetics*. Wiley-Blackwell.
- **8.** Doudna, J. (2017). A crack in creation: The new power to control evolution. Vintage Digital.

Scheme of Examination: The students shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered in	Time allotted for	% Weightage			
Subjective Test	the examination	the examination	(Marks)			
Test I (after 30 days)	20%	1:30 hour	10+10 (20)			
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)			
Major test (after 90 days)	100%	3 hours	60			
Total			100			
Practicals/ Research	Practicals/ Research					
Internal Examination	100%	2 hours	25			
External examination	100%	2 hours	25			
Total	1	1	50			

Note:

1. Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14 out of 40 will be eligible to re-appear in the Test I and Test II only once.

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

• Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.

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• Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B. In major test there should not be a gap of more than two days in between two tests.

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SEMESTER-III

Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028

Semester Cou	irse No.	Course Title	Credits	Assessment Scheme
III P2H		Basics of Bioinformatics & Biostatistics	•	Minor Test I: 5+5 Minor Test II: 5+5 Major test: 30 Total: 50

COURSE OUTCOMES:

CO1: Students will gain knowledge about the foundational concepts in descriptive and inferential statistics, including measures of central tendency, probability distributions, hypothesis testing, and regression analysis relevant to biological data.

CO2: Familiarize students with core bioinformatics resources, including primary and secondary nucleotide and protein sequence databases, and their classification systems.

CO3: Students will get a comprehensive overview of genome projects (e.g., Human Genome Project, Genome India Project), genome mapping and sequencing techniques, and their applications in modern biology.

Unit I: Bioinformatics and Biological Databases

- **1.1 Introduction to Bioinformatics** Role of the internet in modern biological research; Scope and applications of bioinformatics in life sciences; Overview and classification of biological databases: Primary databases, Secondary databases, Composite database
- **1.2 Nucleotide Sequence Databases-** Structure, content, and access to major nucleotide databases: GenBank (NCBI), EMBL-EBI Nucleotide Sequence Database, DDBJ (DNA Data Bank of Japan).
- **1.3 Protein Sequence Databases-** Key repositories for protein sequence information; SWISS-PROT, TrEMBL, UniProt, PROSITE and Pfam, OWL.
- **1.4 Structural Databases and Classification Systems-** Resources for macromolecular 3D structures: Protein Data Bank (PDB), Molecular Modelling Database (MMDB), Nucleic Acid Database (NDB); Structural classification systems: SCOP (Structural Classification of Proteins), CATH (Class, Architecture, Topology, Homologous superfamily)

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SEMESTER-III

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

Unit II: Statistical Methods and Inference

10hours

- 2.1 Descriptive Statistics and Probability Fundamentals- Measures of central tendency: Mean, median, and mode; Measures of dispersion: Range, variance, standard deviation; Fundamentals of probability: Definitions, types (classical, empirical, and subjective); Combinatorics: Permutations and combinations; basic probability computations.
- 2.2 Probability Distributions and Statistical Hypotheses- Theoretical probability distributions: Binomial, Poisson, and Normal; Introduction to hypothesis testing: Null and alternative hypotheses; Statistical errors: Type I and Type II errors
- **2.3 Inferential Statistical Techniques-** Tests of significance: t-test (independent and paired), chi-square test; Analysis of variance (ANOVA): One-way and two-way ANOVA.
- 2.4 Correlation and Regression Analysis- Simple correlation: Concepts, calculation, and interpretation; Simple linear regression: Estimation of parameters, interpretation; Application and limitations in predictive analysis.

UNIT III: Information Retrieval and Computational Analysis of Biological Databases 10hour

- **3.1 Data Retrieval Systems** SRS (Sequence Retrieval System) for flat-file databases, ENTREZ (NCBI) global search platform, LinkDB for pathway and link-based data retrieval.
- 3.2 Sequence Analysis and Submission Tools- Sequence similarity tools: BLAST, FASTA, CLUSTALW; Sequence submission: BankIt, Sequin, Webin, SAKURA
- 3.3 Genomics and the Human Genome Project- Overview of the Human Genome Project, latest advancements: T2T consortium, Human Pangenome Reference Consortium (HPRC), Genome India Project (GIP). Genome sequencing and mapping techniques, Applications of genome maps, Telomere-to-Telomere (T2T) Consortium, Human Pangenome Reference Consortium (HPRC), Genome India Project (GIP), European Reference Genome Atlas (ERGA), Earth BioGenome Project (EBP), Human Heredity and Health in Africa (H3Africa).

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(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

3.4 Genome and Phylogenetic Analysis- Sequence assembly and genome annotation, Phylogenetic analysis methods; Comparative genomics: COGs and HomoloGene (NCBI).

Books Recommended:

- 1. Lesk, A. M. (2019). *Introduction to bioinformatics* (5th ed.). Oxford University Press
- **2.** Ghosh, Z., & Mallick, B. (2008). *Bioinformatics: Principles and applications* (1st ed.). Oxford University Press.
- **3.** Rastogi, S. C., Rastogi, P., & Mendiratta, N. (2022). *Bioinformatics: Methods and applications Genomics, proteomics and drug discovery* (5th ed.). PHI Learning Pvt. Ltd
- **4.** Mount, D. W. (2001). Bioinformatics: Sequence and Genome Analysis.Cold Spring Harbor, NY: Cold Spring Harbor.
- **5.** Baxevanis, A. D., & Ouellette, B. F. F. (2005). *Bioinformatics: A practical guide to the analysis of genes and proteins* (3rd ed.). Wiley-Interscience
- **6.** Pevsner, J. (2015). Bioinformatics and Functional Genomics. Hoboken, NJ.: Wiley-Blackwell.
- 7. Bourne, P. E., & Gu, J. (2009). Structural Bioinformatics. Hoboken, NJ: Wiley-Liss.
- **8.** Lesk, A. M. (2019). *Introduction to protein science: Architecture, function, and genomics* (3rd ed.). Oxford University Press.
- **9.** Attwood, T. K., & Parry-Smith, D. J. (2009). *Introduction to bioinformatics* (2nd ed.). Pearson Education Limited
- **10.** Information Resources Management Association. (Ed.). (2024). *Research anthology on bioinformatics, genomics, and computational biology* (Vols. 1-3). IGI Global.

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(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

Scheme of Examination: The student shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered	Time allotted for the	% Weightage
Subjective Test	in the examination	examination	(Marks)
Test I (after 30 days)	20%	1 hour	05+05 (10)
Test II (after 30 days)	21-40%	1 hour	05+05 (10)
Major test (after 90 days)	100%	2.5 hours	30
	Total		50
	Practical/Resea	arch	
Internal Examination	100%	2hours	25
External Examination	100%	2hours	25
Total	•		50

Note:

Major test will have two sections (A & B). Section A is compulsory comprising of 10 questions of 1 mark each and be spread over entire syllabus. Section B comprises of 4 questions from remaining 2 units and candidate must attempt one question from each unit of 10 marks each.

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SEMESTER-III

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

Semester	Course No.	Course Title	Credits	Assessment Scheme
III	P2HGTC304	Fundamentals of Clinical Genetics	Theory: 04	Minor Test I: 10+10 Minor Test II: 10+10 Major test: 60 Total: 100

COURSE OUTCOMES:

CO1: Students will understand the interplay between genetic and environmental factors in the development of clinical disorders and assess genetic risk using tools like polygenic risk scores.

CO2: Students will recognize the genetic basis, inheritance patterns, and clinical features of monogenic and multifactorial disorders, including both autosomal and X-linked conditions.

CO3: Students will analyze complex diseases with polygenic and environmental components, such as cardiovascular, neurodegenerative, and psychiatric disorders, to inform diagnosis and management.

CO4: Students will be able to explore the role of dermatoglyphics in identifying genetic disorders and its clinical applications.

CO5: Students will understand the diagnosis and genetic basis of congenital anomalies and the use of emerging reproductive technologies.

UNIT I: Genetics, Environment & Disease

12hours

- **1.1** Genetic and Environmental determinants of Clinical Disorders.
- 1.2 Clinical utilization of presymptomatic and predisposition testing for genetic diseases and malignancy.
- **1.3** Nature and Nurture: Disentangling the effects of genes and environment.
- **1.4** Risk of recurrence, empiric risks and polygenic risk scores.

UNIT II Monogenic/Unifactorial Genetic Disorders

12hours

2.1 Introduction to monogenic genetic diseases.

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- **2.2** Common Autosomal Disorders: Thalassemia, cystic fibrosis, lysosomal storage diseases, Neurofibromatosis, Achondroplasia.
- **2.3** Common X-linked Disorders: Hemophilia, DMD, G-6-PD Deficiency.
- **2.4** Disorders with multiple modes of inheritance: Retinitis Pigmentosa, Alport Syndrome, Prelingual deafness.

UNIT III: Polygenic/Multifactorial Disorders

12hours

- **3.1** Introduction to complex genetic disorders.
- 3.2 Coronary Artery Disease and MIYA (Myocardial Infarction in young adults).
- 3.3 Inflammatory Bowel Disease (IBD): Crohn's Disease and Ulcerative Colitis.
- **3.4** Neurodegenerative & Psychiatric disorders: Parkinson, Alzheimer's, Schizophrenia & bipolar disorder.

UNIT IV: Dermatoglyphics in Clinical Genetics

12hours

- **4.1** Introduction to dermatoglyphics
- **4.2** Classification and pattern types
- **4.3** Dermatoglyphics in clinical disorders- Down's syndrome, Turner's syndrome, Klinefelter's syndrome and Cri du chat syndrome.
- **4.4** Clinical applications, Advantages and Limitations.

UNIT V: Clinical and Genetic Approaches to Congenital Disorders 12hours

- **5.1**Clinical Approach to Children with Congenital Anomalies: Down syndrome, Achondroplasia, Marfan syndrome.
- **5.2** Dysmorphology and Clinical Teratology.
- **5.3** Ring chromosome and related genetic disorders.
- **5.4** Reprogenetics- Germinal Choice Technology.

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(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

Books Recommended:

- **1.** Pyeritz, R. E., Korf, B. R., & Grody, W. W. (Eds.). (2024). Emery and Rimoin's principles and practice of medical genetics and genomics (8th ed.). Academic Press.
- **2.** Turnpenny, P. D., Ellard, S., & Cleaver, R. (2021). Emery's elements of medical genetics and genomics (16th ed.). Elsevier.
- **3.** D. Peter Snustad and Michael J.Simmons. Principles of Genetics. 6th edition. John Wiley & Sons, Inc., 2011.
- **4.** Kingston, H. M. (2002). ABC of clinical genetics (3rd ed.). BMJ Books.
- **5.** Human Heredity: Principles and Issues by Micheal R. Cummings; 11th edition, Cengage Learning, 2016.

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MCQ on LMS+	Syllabus to be covered	Time allotted for the	% Weightage
Subjective Test	in the examination	examination	(Marks)
Test I (after 30 days)	20%	1:30 hour	10+10 (20)
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)
Major test (after 90 days)	100%	3 hours	60
	100		

Note:

1. Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14 out of 40 will be eligible to re-appear in the Test I and Test II only once.

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

• Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.

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• Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B. In major test there should not be a gap of more than two days in between two test

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SEMESTER-III

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

Semester	Course No.	Course Title	Credits	Assessment Scheme
Ш	P2HGTE308	Stem Cell Technology	Theory: 02 Practical: 02 Total: 04	Minor Test I: 5+5 Minor Test II: 5+5 Major test: 30 Total:50

COURSE OUTCOMES:

CO1: Students will be able to learn the fundamental concepts of stem cell biology, including stemness, types of stem cells, their sources, and laboratory techniques for isolation, characterization, and maintenance.

CO2: Students will explore the therapeutic potential and applications of mesenchymal and induced pluripotent stem cells (iPSCs) in regenerative medicine and disease modeling.

CO3: Students will acquire understanding of the ethical, legal, and regulatory frameworks governing stem cell research, including national and international guidelines and the role of biobanks in clinical and research settings.

UNIT I: Introduction to Stem Cells

10hours

- **1.1** Introduction to stem cells and basis of stemness
- **1.2** Types of stem cells: Embryonic stem cells, adult stem cells, hematopoietic stem cells, mesenchymal stem cells, cancer stem cells, induced pluripotent stem cells, Placental cell and umbilical cord stem cells, adipocyte derived stem cells
- **1.3** Isolation, characterization, and maintenance of embryonic stem cells.
- **1.4** Serum and feeder free culture of human embryonic stem cells, cryopreservation of embryos.

UNIT II: Mesenchymal & Induced Pluripotent Stem Cells

- **2.1** Introduction to mesenchymal stem cells, isolation, and characterization.
- **2.2** Differentiation of mesenchymal stem cells into various lineages.
- **2.3** Introduction to iPSC technology; Reprogramming iPSCs: integration and non-integration methods;
- **2.4** Advantages and disadvantages of iPSCs.

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SEMESTER-III

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UNIT III: Stem Cell Applications & Ethical Issues

10hours

- **3.1** Existing and potential clinical use of stem cells.
- **3.2** Application of stem cells to regenerative medicine.
- **3.3** Biobanks &Controversies in stem cell research, current controversies surrounding stem cell research.
- **3.4** Ethical and regulatory issues associated with stem cell research: National and International guidelines.

Books Recommended:

- 1. Lanza, R., & Atala, A. (Eds.). (2025). *Essentials of stem cell biology* (4th ed.). Elsevier Science.
- **2.** Brand-Saberi, B. (Ed.). (2020). Essential current concepts in stem cell biology. Springer Nature.
- **3.** Deb, K. D., &Totey, S. M. (Eds.). (2011). *Stem cell basics and application*. Tata McGraw-Hill Pvt. Ltd.
- 4. Chiu, A. Y., & Rao, M. (Eds.). (2011). Human embryonic stem cells. Humana Press.
- 5. Sell, S. (Ed.). (2010). Stem cells handbook. Humana Press.
- 6. Marshak, D. R., Gardner, R. L., & Gottlieb, D. I. (2001). Stem cell biology.
- 7. Lanza, R., Gearhart, J., & Hogan, B. (Eds.). Essentials of stem cell biology.

Scheme of Examination: The student shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered	Time allotted for the	% Weightage		
Subjective Test	in the examination	examination	(Marks)		
Test I (after 30 days)	20%	1 hour	05+05 (10)		
Test II (after 30 days)	21-40%	1 hour	05+05 (10)		
Major test (after 90 days)	100%	2.5 hours	30		
	50				
Practical/Research					
Internal Examination	100%	2hours	25		
External Examination	100%	2hours	25		
Total			50		

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SEMESTER-III

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Note: Major test will have two sections (A & B). Section A is compulsory comprising of 10 questions of 1 mark each and be spread over entire syllabus. Section B comprises of 4 questions from remaining 2 units and candidate has to attempt one question from each unit of 10 marks each.

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SEMESTER-III

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028

Semester	Course No.	Course Title	Credits	Assessment Scheme
Ш	P2HGTE309	Cancer Biology	Theory: 02 Practical: 02 Total: 04	Minor Test I: 5+5 Minor Test II: 5+5 Major test: 30 Total:50

COURSE OUTCOMES:

CO1: Students will acquire foundational knowledge of cancer biology, including cancer types, stages, and hallmark characteristics, with emphasis on molecular and cellular mechanisms of oncogenesis.

CO2: Students will be able to analyze genetic, epigenetic, and environmental factors contributing to cancer development, including tumor suppressor genes, oncogenes, chromosomal instability, and the tumor microenvironment.

CO3: Familiarize students with modern approaches to cancer diagnosis, treatment, and genetic counseling, including personalized therapies, immunotherapy, liquid biopsies, and polygenic risk assessment following established guidelines (e.g., MCC – Multidisciplinary Cancer Care Guidelines).

Unit 1: Fundamentals of Cancer Biology

10hours

- **1.1** Definition and overview of cancers, Hallmarks of cancer cells.
- 1.2 Types of Cancers: Benign vs. malignant tumors; Carcinomas Sarcomas, Leukemias, Lymphomas & Myelomas, Hematological & Solid, Inherited & Familial
- 1.3 Stages of Cancer Development: Initiation, promotion and progression & Clonal evolution.
- **1.4** Cancer risk factors: Lifestyle choices, environmental exposures, and genetic predisposition, molecular mechanisms of carcinogens & mutagens

Unit II: Molecular and Cellular Mechanisms of Cancer

- 2.1 Molecular Basis of Cancer: Two-hit hypothesis, Tumor suppressor genes and Oncogenes; Chromosomal Instability in Oncogenesis
- **2.2** The Tumor Microenvironment: Driving Cancer Progression.

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- **2.3** Epigenetics in Cancer: DNA methylation, histone modifications, non-coding RNAs (ncRNAs) & chromatin remodelling.
- **2.4** Role of Cell Cycle in Carcinogenesis.

Unit III: Managing Cancer: From Heredity to Therapy

10hours

- **3.1** Familial cancer syndromes: BRCA1/2 breast and Ovarian cancer syndromes; Lynch syndrome (HNPCC), Li-Fraumeni syndrome; Familial adenomatous polyposis (FAP).
- **3.2** Detection and Diagnosis: Biopsy techniquesand types; Imaging modalities, Tumor markers & monoclonal antibodies.
- **3.3** Cancer therapies: chemotherapy; Immunotherapy and targeted therapies (with special emphasis on CAR T Therapy) & Cancer vaccines.
- **3.4** Genetic Counselling and Polygenic risk assessment for cancer predisposition, Multidisciplinary Cancer Care (MCC) Guidelines.

Recommended Books:

- **1.** Cohn, R. D., Scherer, S. W., &Hamosh, A. (Eds.). (2024). *Thompson & Thompson genetics and genomics in medicine* (9th ed.). Elsevier.
- 2. Weinberg, R. A. (2023). The biology of cancer (3rd ed.). W. W. Norton & Company.
- **3.** Hesketh, R. (2023). *Introduction to cancer biology* (2nd ed.). Cambridge University Press.
- **4.** Pecorino, L. (2021). *Molecular biology of cancer: Mechanisms, targets, and therapeutics* (5th ed.). Oxford University Press.
- **5.** DeVita, V. T., Hellman, S., & Rosenberg, S. A. (2019). *Cancer: Principles and practice of oncology* (9th ed.). Wolters Kluwer Health.
- **6.** Heim, S., & Mitelman, F. (Eds.). (2015). *Cancer cytogenetics: Chromosomal and molecular genetic aberrations of tumor cells* (4th ed.). Wiley-Blackwell.

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SEMESTER-IV

(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

Scheme of Examination: The student shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered	Time allotted for the	% Weightage		
Subjective Test	in the examination	examination	(Marks)		
Test I (after 30 days)	20%	1 hour	05+05 (10)		
Test II (after 30 days)	21-40%	1 hour	05+05 (10)		
Major test (after 90 days)	100%	2.5 hours	30		
	50				
Practical/Research					
Internal Examination	100%	2hours	25		
External Examination	100%	2hours	25		
Total	•		50		

Note:

Major test will have two sections (A & B). Section A is compulsory comprising of 10 questions of 1 mark each and be spread over entire syllabus. Section B comprises of 4 questions from remaining 2 units and candidate has to attempt one question from each unit of 10 marks each.

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(Syllabi for the examinations to be held in the years Dec. 2026, 2027, 2028)

COURSE CODE	COURSE NAME	CREDITS				
	CREDIT FRAMEWORK FOR SEMESTER-IV					
	Major Core [8 (T)+ 4 (P)]					
P2HGTC401	Essentials of Genetic Counseling	4				
P2HGTC402	Fundamentals of Immunology	4				
P2HGPC403	Practical based on P2HGTC401	2				
P2HGPC404	Practical based on P2HGTC402	2				
Total Credits 12						
Major Elective (any one* 2T)						
P2HGTE405	Basics of Artificial Intelligence	2*				
P2HGTE406	Bioethics and IPR	2				
Tota	2					
P2HGRC407	Research Project +	16				
Presentation/Dissertation						
,	30					

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Semester	Course No.	Course Title	Credits	Assessment Scheme
IV	P2HGTC401	Essentials of Genetic Counseling	•	Minor Test I: 10+10 Minor Test II: 10+10 Major test: 60 Total: 100

COURSE OUTCOMES:

- **CO1**: Students will gain foundational knowledge of genetic counseling, including its types, process, and significance in Mendelian and non-Mendelian disorders.
- **CO2**: Students will explore the psychosocial, ethical, and behavioural dimensions of counseling for individuals, families, and groups affected by genetic conditions.
- **CO3**: Students will examine genetic counseling approaches in specific disorders, such as neuromuscular diseases, infertility, hereditary cancers, and developmental anomalies.
- **CO4**: Students will understand national and international ethical guidelines, informed consent, genetic discrimination, and the functioning of genetic counselling clinics.
- **CO5**: Students will be familiarized with legal frameworks, including the PCPNDT Act, regulations on genetic services, and the legal responsibilities of genetic counsellors.

Unit-I: Introduction to Genetic Counseling

12hours

- **1.1** An overview of the history and development of Genetic Counseling
- **1.2** The fundamental components and skills utilized in pediatric and adult genetic counseling.
- **1.3** Types of Genetic Counseling: Individual, Family and Group.
- **1.4** Indications of Genetic counseling in prenatal, pediatric and adult-onset genetic disorders.

Unit-II: The Genetic Counseling Process

- **2.1** Taking clinical/medical history, family history and its interpretation, Bayesian theorem.
- **2.2** Psychosocial and Behavioral aspects of Genetic Counseling.
- **2.3** Explaining risks and reproductive options in cytogenetic disorders, Mendelian and Non-Mendelian disorders.

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2.4 Understanding the Genetic Counseling context: Screening verses Diagnostics testing

Unit-III Genetic Counseling in Genetic Disorders

12hours

- **3.1** Neuromuscular Disorders/Deafness and Blindness.
- **3.2** Infertility and Recurrent Pregnancy Loss.
- **3.3** Hereditary and Familial Cancers.
- **3.4** Trinucleotide repeat expansions: Understanding anticipation.

Unit-IV Fundamentals of Pre-test and post-test Counseling

12hours

- **4.1** Explaining the advantages and limitations of tests and interpretation results of the genetic tests (Chromosomal, biochemical, and molecular diagnostic tests).
- **4.2** Determining the mode of inheritance and risk of occurrenceand recurrence of the genetic condition/birth defects, and appropriate communication.
- **4.3** Explaining the diagnosis, etiology, natural history, monitoring, and management of the genetic disorder and providing counseling for reproductive options.
- **4.4** Providing written documentation of medical, genetic, and counseling information for families (extended screening andcascade testing) and other health professionals.

Unit-V Ethos of Genetic Counseling

12hours

- **5.1** Ethical principles of Genetic Counseling: Privacy, Confidentiality and Genetic Discrimination, Informed Consent and Right of Choice.
- **5.2** Genetic Counseling in Clinics: Types, working & Maintenance of Genetic Registers, preparing Genetic Counseling Reports.
- **5.3** Role of Genetic Counselors in era of Clinical Genomics.
- **5.4** Status of Genetic Counseling in India and Global Scenario.

Books Recommended

- **1.** Pyeritz, R. E., Korf, B. R., & Grody, W. W. (Eds.). (2024). *Emery and Rimoin's principles and practice of medical genetics and genomics* (7th ed.). Academic Press.
- **2.** Cohn, R. D., Scherer, S. W., &Hamosh, A. (Eds.). (2024). *Thompson & Thompson genetics and genomics in medicine* (9th ed.). Elsevier.

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- **3.** Dave, U., & Shetty, D. (2021). *Genetic counseling: Clinical and laboratory approach* (1st ed.). Springer.
- **4.** Veach, P. M., LeRoy, B. S., & Callanan, N. P. (2020). *Genetic counselling practice: Advanced concepts and skills* (2nd ed.). Springer.
- **5.** Clarke, A., Murray, A., & Sampson, J. (2020). *Harper's practical genetic counselling* (8th ed.). CRC Press.
- **6.** Hurst, J. A., & Firth, H. V. (2017). *Oxford desk reference: Clinical genetics and genomics*. Oxford University Press.

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Subjective Test	the examination	the examination	(Marks)
Test I (after 30 days)	20%	1:30 hour	10+10 (20)
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)
Major test (after 90 days)	100%	3 hours	60
Total			100
Practicals/ Research			
Internal Examination	100%	2 hours	25
External examination	100%	2 hours	25
Total	1		50

Note:

1. Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14 out of 40 will be eligible to re-appear in the Test I and Test II only once.

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

• Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.

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• Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B. In major test there should not be a gap of more than two days in between two tests.

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Semester	Course No.	Course Title	Credits	Assessment Scheme
IV	P2HGTC402	Fundamentals of	Theory: 04	Minor Test I: 10+10
		Immunology	Practical:02 Total: 06	Minor Test II: 10+10 Major test: 60
				Total:100

COURSE OUTCOMES:

CO1: Students will be able to explain the organization and function of the immune system, including the roles of various immune cells, antigens, antibodies, and the molecular basis of immune recognition and response.

CO2: Students will develop skill to demonstrate mechanisms underlying humoral and cell-mediated immunity, antigen presentation, complement activation, and cytokine function in immune regulation.

CO3: Students will be able to analyze immunological dysfunctions such as hypersensitivity reactions, autoimmune diseases, immunodeficiency disorders, and graft rejection, including their cellular and molecular basis.

CO4: Students will be able to demonstrate immunological techniques such as ELISA, immunoblotting, flow cytometry, and immunoelectrophoresis for the diagnosis of diseases and characterization of immune responses.

CO5: Students will acquire knowledge to evaluate modern immunotherapies, including CAR-T cell therapy, monoclonal antibodies, and vaccine strategies, and assess their clinical applications and mechanisms of action.

UNIT-I: Introduction to The Immune System

12hours

- 1.1 Introduction to immune system, Innate and acquired immunity, clonal nature of immune response; Organization and structure of lymphoid organs, Hematopoiesisand differentiation, Cells of the immune system: B- lymphocytes, T-lymphocytes, Macrophages, Dentritic cells, Natural killer and Lymphokine activated killer cells, Eosinophils, Neutrophils and Mast cells.
- **1.2** Nature and Biology of antigens and super antigens,
- **1.3** Antibody structure and function, antibody mediated effector functions, antibody classes and biological activity.

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1.4 Antigenic determinants on immunoglobulins, Immunoglobulin superfamily, BCR &TCR, generation of antibody diversity.

UNIT-II: Humoral and Cell Mediated Immunity

12hours

- **2.1** Regulation of immune response, Antigen processing and presentation, generation of humoral and cell mediated immune responses, Activation of B- and T- lymphocytes,
- **2.2** Complement System: components of complement, complement activation, complement cascade, regulation of complement System.
- **2.3** Cytokines, cytokines receptors, cytokines antagonists, role of cytokines in T H 1/T H 2 subset development and their role in immune regulation, MHC: MHC molecules and genes, MHC restriction.
- **2.4** Cell-mediated cytotoxicity: Mechanism of T cell and NK cell mediated lysis, Antibody dependent cell mediated cytotoxicity, macrophage mediated cytotoxicity.

UNITIII: Errors of the Immune System

12hours

- **3.1** Immunological tolerance: central tolerance, peripheral tolerance, component of peripheral tolerance.
- **3.2** Hypersensitivity: IgE mediated Hypersensitivity, Antibody mediated cytotoxic Hypersensitivity, Immune complex- mediated Hypersensitivity, Delayed type Hypersensitivity.
- **3.3** Inborn Errors of Immune System: organ specific and systemic autoimmune diseases, animal models for autoimmune diseases and the molecular mechanism, immunodeficiency disorder- AIDS.
- **3.4** Transplantation immunology: Immunological basis of graft rejection, clinical manifestation of graft rejection, general immunosuppressive therapy, specific immunosuppressive therapy, immune tolerance to allografts.

UNIT IV: Immunodiagnostic Procedures

12hours

4.1 Antigen- Antibody interactions and Techniques – ELISA and its variants, ELISPOT, Radio immunoassay, Immunofluorescence, Flow cytometry and Fluorescence, Immuno electron microscopy.

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- **4.2** Agglutination and haemagglutination assays.
- **4.3** Types of immunodiffusion and immunoelectrophoretic procedures, isoelectric focusing
- **4.4** Affinity chromatographic methods and Immunoblotting.

UNIT V: Immunology-Based Therapies

12hours

- **5.1** Immunotherapy: CAR T-cell therapy, Immune checkpoint inhibitors, Monoclonal antibodies, interleukin therapy, vaccines.
- **5.2** Chimeric antigen receptor (CAR) T-cell therapy: working, application and side effects, approved CAR T-cell therapies.
- **5.3** Hybridoma Technology and Monoclonal antibodies detection and application of monoclonal antibodies.
- 5.4 Vaccines: History of vaccine development, introduction to the concept of vaccine, Active and passive immunization, Designing vaccines for active immunization: Conventional vaccines, subunit vaccines, conjugate vaccines, DNA vaccines, Recombinant vector vaccines.

BOOKS RECOMMENDED

- 1. Punt, J., Stranford, S., Snell, P. J., & Williams, P. J. (2023). *Kuby immunology* (9th ed.). W.H. Freeman and Company.
- **2.** Abbas, A. K., Lichtman, A. H., Pillai, S., & Henrickson, S. (2025). *Cellular and molecular immunology* (11th ed.). Elsevier.
- **3.** Flajnik, M. F., Singh, N. J., & Holland, S. M. (Eds.). (2022). *Paul's fundamental immunology* (8th ed.). Wolters Kluwer Health.
- 4. Immunology, International 9th Edition 2020 by David Male.
- 5. Coleman, R.M., Lombard, M.F. and Sicard, R.E.(1992). Fundamental Immunology.
- **6.** Male, D., Brostoff, J., Roth, D., &Roitt, I. (Eds.). (2012). *Immunology* (8th ed.). Elsevier Health Sciences
- 7. Benjamini, E., Coico, R., & Sunshine, G. (2021). *Immunology: A short course* (8th ed.). Wiley-Blackwell
- 8. Davies, H. (2024). Introductory immunobiology. CRC Pres.
- 9. Wood, P. (2011). Understanding immunology (3rd ed.). Pearson Education.

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Scheme of Examination: The students shall be continuously evaluated during the conduct of each course based on his/her performance as follows:

MCQ on LMS+	Syllabus to be covered in	Time allotted for	% Weightage
Subjective Test	ective Test the examination the examination		(Marks)
Test I (after 30 days)	20%	1:30 hour	10+10 (20)
Test II (after 30 days)	21-40%	1:30 hour	10+10 (20)
Major test (after 90 days)	100%	3 hours	60
Total			100
Practical's/ Research			
Internal Examination	100%	2 hours	25
External examination	100%	2 hours	25
Total			50

Note:

1. Test I and Test II

The Subjective Test of Test I and Test II would consists of three short answer type questions (05marks each). Students are required to answer any two questions. No preparatory holidays shall be provided for the Test I and Test II. Those candidates who have appeared in Test I and Test II and failed to get the minimum required marks i.e. 14 out of 40 will be eligible to re-appear in the Test I and Test II only once.

2. Major test

The Major test will comprise of two sections, Section- A and Section- B.

- Section A will have one compulsory question comprising of 08 parts (minimum 01 from each unit) of 03 marks each.
- Section B will have 06 questions of 12 marks each to be set from the last three units (02 from each unit). Students are required to attempt 01 question from each unit of section B.
 In major test there should not be a gap of more than two days in between two tests.

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SEMESTER-IV

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Semester	Course No.	Course Title		Credits	Assessment Scheme
IV	P2HGTE405	Basics of Intelligence	Artificial	Theory: 02	Minor Test I: 5+5 Minor Test II: 5+5 Major test: 30 Total: 50

COURSE OUTCOMES:

CO1: Students will be able to introduce the fundamentals of Artificial Intelligence and biological data, including types of data, data processing techniques, and ethical concerns such as privacy and algorithmic bias.

CO2:Students will be able to explore core machine learning and deep learning techniques, including supervised, unsupervised, and reinforcement learning, with practical applications in biological research using tools like BioPython and AlphaFold.

CO3: Students will be able to demonstrate real-world applications of AI in biology, such as drug discovery, disease diagnosis, crop improvement, and environmental sustainability, highlighting ethical considerations in AI-driven biological research.

UNIT I: Introduction to AI and Databases

10hours

- **1.1 Artificial intelligence and Its foundations** Definition and history of AI; Branches of AI: Machine Learning (ML), Deep Learning (DL): AI vs. traditional programming: Generative AI: ChatGPT, DeepSeek: AI vs. Biological Intelligence.
- **1.2 Biological data**: Types of biological data: imaging, experimental, clinical, and environmental: Challenges in biological data analysis.
- **1.3 Computational tools and data management**, Need for computational tools in biology: Data storage and analysis: Cloud vs. server-based data storage and analysis: Data privacy and security: Algorithmic bias and fairness: Sources of bias in biological datasets.
- **1.4 Data processing techniques**: Data cleaning and handling: Dealing with missing values, noise, and outliers: Data transformation: Normalization, standardization, and scaling of data.

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UNIT II: Machine Learning and Deep Learning

10hours

- **2.1** Machine learning; Types of ML: Supervised, unsupervised, Reinforcement learning; Use cases in biology: AlphaFold, trRosetta, AIDDISON; Common tools and libraries: Bio Python, EMBOSS, etc.
- **2.2** Deep Learning architectures, Convolutional Neural Networks (CNNs) and Recurrent Neural Networks (RNNs): Applications, advantages, and limitations in biology.
- **2.3 Data processing techniques**: Data cleaning and handling: Dealing with missing values, noise, and outliers: Data transformation: Normalization, standardization and scaling of data.
- **2.4 Programming languages:** Python, Bio Python, R, Shell/Bash; Practical examples and hands-on coding exercises. Programming without coding.

UNIT III: Applications in Biology

10hours

- **3.1 Bioinformatics:** Overview of commonly used AI-based tools, AI-based phenotype tools and databases for nucleic acid protein, metabolite analysis.
- **3.2 Drug discovery and development**: Target identification and validation: Lead discovery and optimization: Preclinical and clinical trial data analysis.
- **3.3 Use of AI in Human Diseases:** Cancer Diagnosis, Imaging and Pattern Recognition in Syndromes(AI tools: Face2Gene and Gestalt Matcher). Drug repurposing using molecular and clinical data (AI tools: DeepChem, DeepTox, ChEMBL etc.).
- **3.4 Agricultural and environment:** Crop yield prediction; Disease and pest detection; Resource optimization (e.g., water, fertilizers); Environmental monitoring and sustainability. Ethical principles and bias in AI applications for biology.

Books Recommended:

- **1.** Raja Sekar, M. N., Saxena, A., et al. (2021). Artificial Intelligence in Precision Health: From Concept to Practice. Springer.
- **2.** Bennett, M. (2025). A Brief History of Intelligence: Why the Evolution of the Brain Holds the Key to the Future of AI.
- **3.** Olson, P. (2024). *Supremacy: AI, ChatGPT and the Race That Will Change the World.* Pan Macmillan/St. Martin's Press.

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- **4.** Birney, E., Durbin, R. M., et al. (2020). Genomic Data Science and Its Practical Applications. Academic Press
- 5. Wold, B. J. (2019). Databases and Data Management in Human Genetics. Elsevier.

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Subjective Test	in the examination	examination	(Marks)
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Major test (after 90 days)	100%	2.5 hours	30
Total			50

Note:

Major test will have two sections (A & B). Section A is compulsory comprising of 10 questions of 1 mark each and be spread over entire syllabus. Section B comprises of 4 questions from remaining 2 units and candidate must attempt one question from ach unit of 10 marks each.

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Semester	Course No.	Course Title	Credits	Assessment Scheme
IV	P2HGTE 406	Bioethics and IPR	Theory: 02	Minor Test I: 05+05 Minor Test II: 05+05 Major test: 30 Total:50

COURSE OUTCOMES:

CO1: Students will be able tofamiliarize with the fundamentals of Intellectual Property Rights (IPRs) and their specific applications in biotechnology, including patents, trademarks, copyrights, and sui generis protections.

CO2: Students will be able tounderstand the procedures and challenges of patenting in the life sciences, including patent filing, licensing, technology transfer, and the roles of national and international patent authorities.

CO3: Students will be able to critically examine ethical and legal issues in biotechnology, including gene therapy, GMOs, organ transplantation, and environmental protection, guided by national policies and international bioethical principles.

UNIT-I: Intellectual Property

10hours

- **1.1** Introduction to IPRs; various types of IPRs: Trademarks, Copyrights, Geographical indications, Trade Secrets; Role of IPRs in Biotechnology
- **1.2** Patents: Criteria for patenting in Biotechnology/Microbiology/Biochemistry: novelty, non-obviousness, and utility, patentable and non-patentable inventions; Biological Patents; Purpose of patents.
- **1.3** *Sui generis* system of IPRs: Need for *Sui generis* system; Plant variety protection, Database protection, other forms of *Sui generis* protection.
- **1.4** Introduction to WIPO and TRIPS, Various provisions in the TRIPS Agreement; Indian legislations for the protection of various types of IPs; National Biodiversity protection initiatives.

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UNIT-II: Patenting 10hours

- **2.1** Specific challenges in biological patenting: gene patents, diagnostic patents, and biopharmaceuticals patents.
- **2.2** Patent application: various components of Patent application, patent search, patent filing, Pre grant and post grant opposition, Patent Cooperation Treaty (PCT).
- 2.3 Introduction to Indian patent office, US patent office and European patent office.
- **2.4** Patent Licensing; Technology Transfer; The role of technology transfer offices in academic and research institutions; Patent Infringement.

UNIT-III: Bioethics 10hours

- **3.1** Traditional knowledge and bioethics, Ethical Issues, Statement of Bioethical Principles.
- **3.2** Gene Therapy, Germ line Gene therapy Moratorium; Medical privacy and genetic discrimination; Organ transplantation.
- **3.3** Bioethics in research: stem cells, animal cloning; Use of animals in research, animal rights; Human experimentation, Medico Legal issues.
- **3.4** Genetically Modified foods, environmental risk, labelling and public opinion; Protection of environment and biodiversity, biopiracy.

Books recommended:

- 1. Philippe Cullet; (2005) Intellectual Property Rights and Sustainable Development, Lexis, Nexis, New Delhi.
- 2. Arthur William et al.; (2005) Expanding Horizons in Bioethics, Springer.
- **3.** Prabuddha Ganguli; (2001) Intellectual Property Rights, Unleashing the Knowledge Economy, Tata Mc Graw Hill, New Delhi.
- 4. Venkatratnam J.B (2009) Entrepreneurship Development, Heritage Printers, Hyderabad
- 5. Patzelt, Holger; Brenner, Thomas (2008), Handbook of Bioentrepreneurship, Springer Publications
- **6.** S. Mitra; (2009) Entrepreneur Journeys Vol. 1, Hachette Publishing Group.
- 7. R.D. Hisrich; (2006) Entrepreneurship, Tata McGraw-Hill Publishing Co Ltd, 6th Edition.
- **8.** D.F. Kuratko, H.P. Welsch; (2001) Strategic Entrepreneurial Growth, Harcourt College Publishers.
- **9.** P. Ganguli; (2009) Intellectual Property Rights, Tata Mcgraw Hill Publishing Co Ltd.M.B. Rao; (2008) Biotechnology, IPRs and biodiversity, Pearson Publications

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