INSTITUTE OF HUMAN GENETICS

M. Sc. Human Genetics

Program Outcome (PO)

PO-1 **Knowledge:** The degree in M.Sc. Human Genetics offers knowledge about various aspects and concepts of Human genome and related genetic disorders.

PO-2 **Skills:** The course offers unique skills and exposure to the students about the most relevant techniques used for the genetic testing purposes and emphasizes for making genetic testing relevant in day to day life.

PO-3 **Usage:** The course provides suitable platform to use different genetic tools for the proper detection and diagnosis of both common and complex genetic disorders. The course also stresses upon the use of genetic concepts for generating awareness among general public. Surveys can be conducted regarding the identification of various genetic disorders.

PO-4 **Analytical based studies with result outcome:** After understanding principles of genetics & practical exposure one can identify the high risk and low risk individuals for a particular genetic trait. Based on the survey conducted and literature studied, students can identify there levant researchable subjects; make concrete proposals with definite outcomes as per defined objectives considering practical aspects of Human Genetics.

PO-5 **Formulation of projects and research there of:** On the basis of the research information, students can design their unique research proposals. Relevant genetic experimentations and investigations can be done. The results can be implemented to improve the diagnosis and treatment of diseases. The data generated can be added to the various databases.

PO-6 **Rural population and Human Genetics:** The prime focus of the course is to promote health by understanding the genetic basis of common diseases and

early detection of potentially treatable genetic conditions. The awareness inthe society about these problems is necessary for improving human health in rural population. The students can trace the genetic etiology of rare genetic diseases in rural areas.

PO -7 Professional Ethics: Application of ethical commitments to follow professional ethics, standards, recommendations and instructions while practicing Human Genetics. The course will be helpful in guiding the students about various special issues in genetic testing viz. Genetic discrimination, Confidentiality, Privacy and PCPNDT Act while performing genetic counseling and diagnosis.

M.Sc. Human Genetics

I[st] Semester (CBCS)

Syllabus for the examination to be held in December, 2022, 2023, 2024

Course No. PSHGTC-101 **Course Title: - Cell Biology**
Credits: 04 **Maximum Marks: 100**
Time Duration: 3.0Hrs **Minor Test I : 20**
Minor Test II : 20
Major Test : 60

Course Outcomes (CO)

CO-1: -The course has been designed to enable the students to understand the structure and function of cell membrane and related transport mechanisms,
CO-2: Students will be able to understand and function of different cell organelles and bimolecular
CO-3: Understanding about the processes of cell division.
CO-4: Knowledge about the mechanism of cellular processes- cell cycle and regulation,
CO-5: Cellular energetic, Signal transduction and Programmed cell death are some of the mechanism which will make a student to have better understanding of the cell.

Unit-I **(13hrs)**

Cell: The basic unit of life

1.1 Cell: Structure and Organization
1.2 Plasma Membrane
1.2.1 Structure of Plasma Membrane with Special emphasis on various models and Functions of Plasma Membrane
1.2.2 Transport across membrane
1.2.3 Mechanisms of Endocytosis and Exocytosis
1.3 Cytoskeleton
1.3.1 Microfilaments: Structural organization, Cell motility and cell shape
1.3.2 Microtubule: Structural and Functional organization, Intermediate filaments
1.3.3 Cilia, Flagella, Centriole

UNIT-II **(12hrs)**

Understanding Cell Organelles

2.1 Mitochondria
2.1.1 Ultrastructure
2.1.2 Role of mitochondria in the formation of ATP
2.2 Endomembrane system
2.2.1 General organization of transport within and outside the cell
2.2.2 Protein sorting and secretion
2.3. Structure and Function:
2.3.1 Nucleus
2.3.2 Ribosomes, Biosynthesis of Ribosomes
2.3.3 Introduction to Golgi complex.
2.3.4 Lysosomes
2.3.5 Peroxisomes

UNIT-III **(12hrs)**

Cell division

3.1 Overview of Cell cycle
3.2 Mitosis
3.2.1 Phases of Mitosis
3.2.2 Significance and Consequences of Mitosis
3.3 Meiosis
3.3.1 Premeiotic and Meiotic stages
3.3.2 Chromosome synapsis and Synaptonemal complex
3.2.3 Mechanism of crossing over, genetic recombination & Meiotic defects
3.2.4 Genetic Control of Meiosis
3.2.5 Genetic Consequences of Meiosis

M.Sc. Human Genetics

I[st] Semester (CBCS)

Syllabus for the examination to be held in December, 2022, 2023, 2024

Course No. PSHGTC-101 | **Course Title: - Cell Biology**
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Major Test : 60

Unit-IV **(13hrs)**

Cellular interactions and Cell Cycle

4.1 Cell-Cell Interaction
- 4.1.1 Cell adhesion molecules
- 4.1.2 Cellular Junctions
- 4.1.3 Extracellular matrix

4.2 Cell cycle and its regulation
- 4.2.1 Cyclin and Cyclin dependent kinases.
- 4.2.2 Regulation of Cyclins, sister Chromatids, Cohesion Remodeling
- 4.2.3 Centrosome Cycle
- 4.2.4 Cell cycle check points
- 4.2.5 Role of Rb and p53 protein in cell cycle regulation.

Unit-V **(12hrs)**

Cell Signalling

5.1 Basic concept of cell signaling (Paracrine, Autocrine, Endocrine, Synaptic, Juxtacrine)
5.2 Intracellular receptor and cell surface receptors
5.3 G-protein linked receptors signalling (via Adenyl cyclase, Phosphatidylinositol effectors)
5.4 Desensitization (termination of GPCR signalling)
5.5 Enzyme linked receptor signalling (RTK signalling; Ras-MAPK signalling; JAK-STAT pathway, Notch and Wnt pathway)
5.6 Nitric oxide Signalling
5.7 Programmed cell death (Apoptosis)

Note for paper setting:

Examination theory weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	upto 20%	1.5 hrs	20
Minor Test-II	21-40%	1.5 hrs	20
Major Test	41-100%	3.0 Hrs.	60

i. Major test will have two sections (A&B)

ii. Section A is compulsory comprising of 10 questions of 1.5 marks each and be spread over entire syllabus

iii. Section B comprises of 6 questions (2 from each unit) from the remaining 3 units and candidate has to attempt one question from each unit (15 marks each).

Books Recommended

1. Alberts et al. Essential Cell Biology, 1998.
2. Purohit S.S Powar. The Cell and the Molecular Biology, 2008,
3. Geoffrey M. Copper and Robert E. Hausman.. The Cell: A Molecular Approach, Eighth Edition. ASM Press and Sinauer Associates, Inc.,2013
4. David Friefelder. Molecular Biology 2013
5. Mousami Debnath,. Cell and Molecular Biology. Shashi Jain Publ. Jaipur, 2014
6. Bruce Alberts et.al. Molecular Biology of the Cell, 6th Edition, Taylor & Francis Group, 2014.
7. Gerald Karp. Cell and Molecular Biology: Concepts and Experiments, 8th Edition John Wiley and Sons, 2016.
8. Rastogi, V.B. Cell Biology. Third Edition. New Age International Publishers, 2016

M.Sc. Human Genetics

I[st] Semester (CBCS)

Syllabus for the examination to be held in December, 2022, 2023, 2024

Course No. PSHGTC-102 **Title: - Human Physiology**
Credits: 04 **Maximum Marks : 100**
Time Duration: 3.0Hrs **Minor Test I : 20**
Minor Test II : 20
Major Test : 60

Course Outcomes: - Course is designed to apprise the student about the details of physiological aspects various organ systems of human body.

CO-1: Understanding about the basic concepts of Human Skeletal system
CO-2: Knowledge about the physiology of digestive system and respiratory system
CO-3: Knowledge about the physiology of cardiovascular system and nervous system
CO-4: Understanding the concepts of Human Endocrinology
CO-5: Understanding the concepts of human fertilization and embryonic development.

Unit-I
Skeletal system **(13 hrs)**

1.1 Bones
- 1.1.1 Classification
- 1.1.2 Histology
- 1.1.3 Ossification
- 1.1.4 Growth
- 1.1.5 Fracture and Repair of bones
- 1.1.6 Joints and their types

1.2 Muscles
- 1.2.1 Classification and structure of different types of muscles.
- 1.2.2 Physiology of muscle contraction
- 1.2.3 Neuromuscular Junction

Unit-II **(12hrs)**
Human Systems and their Physiology

2.1 Gross anatomy of Human Digestive system
- 2.1.1 Physiology of Digestion: in mouth, stomach, Pancreas, Liver, Gall Bladder, Small Intestine, Large Intestine
- 2.1.2 Hormones of Digestive System

2.2 Gross anatomy of Human Respiratory system
- 2.2.1 Physiology of Respiration: Exchange of oxygen and carbon dioxide, Transport of oxygen
- 2.2.2 Control of Respiration
- 2.2.3 Electron Transport Chain

Unit-III
Human Systems and their Physiology–II **(13 hrs)**

3.1 Gross anatomy of Human Cardiovascular system
- 3.1.1 Blood & its Components
- 3.1.2 Anatomy & Physiology of Human heart (Cardiac Cycle, Cardiac output)

3.2 Nervous system
- 3.2.1 Structure of Brain & Spinal Cord
- 3.2.2 Action Potential
- 3.2.3 Neurotransmission

Unit-IV **(13hrs)**
Human Systems and their Physiology -III

4.1 Gross anatomy of Human Excretory system
- 4.1.1 Gross Anatomy of Kidney (structure of Nephron)
- 4.1.2 Physiology of Excretion: Glomerular filtration, Tubular reabsorption, Tubular Secretion, Urine production.

4.2 Physiology of Endocrine System
- 4.2.1 Pituitary Gland

M.Sc. Human Genetics

Ist Semester (CBCS)

Syllabus for the examination to be held in December, 2022, 2023, 2024

Course No. PSHGTC-102 **Title: - Human Physiology**

Credits: 04 **Maximum Marks : 100**

Time Duration: 3.0Hrs **Minor Test I : 20**

Minor Test II : 20

Major Test : 60

4.2.2 Thyroid, Parathyroid Gland

4.2.3 Adrenal Gland

4.2.4 Islets of Langerhans

4.2.5 Gonads

4.2.6 Endocrine Disorders/Metabolic Disorders

Unit-V

Human Embryology **(12 hrs)**

5.1 Male & female Reproductive System

5.2 Mechanism of Human Fertilization

5.3 Establishment of Body Axis

5.4 Implantation

5.5 Development of human embryo upto three germinal layers

5.6 Development of embryonic disc, notochord formation & Neurulation

5.7 Chronic formation & development of placenta

Note for paper setting:

Examination theory weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1.5 Hrs	20
Minor Test-II	21-40%	1.5 Hrs	20
Major Test	41-100%	3 Hrs	60

i. Major test will have two sections (A&B)

ii. Section A is compulsory comprising of 10 questions of 1.5 marks each and be spread over entire syllabus

iii. Section B comprises of 6 questions (2 from each unit) from the remaining 3 units and candidate has to attempt one question from each unit (15 marks each).

Books Recommended:-

1. J. Matthew Neal. How the Endocrine system works, Blackwell Science, 2001
2. Gerard J. Tortora, Principles of Anatomy and Physiology, 2014,.
3. Melmed et al., William's textbook of Endocrinology, 13th edition, 2015.
4. Inderbir Singh, Human Embryology, 11th edition, 2017,
5. GK Pal, Medical Physiology, 13th Edition, Orient Black Swan, 2018.
6. K Sembulingam, Essentials of Medical Physiology, 8th Edition, JAPI Brothers Medical Publishers, 2019.
7. Guyton and Hall. Text book of Medical Physiology. 12th Edition. Elsevier Saunders Publishers, 2019.

M.Sc. Human Genetics

I[st] Semester (CBCS)

Syllabus for the examination to be held in December, 2022, 2023, 2024

Course No. PSHGTC-108 **Course Title: - Human Molecular Biology**
Credits: 04 **Maximum Marks : 100**
Time Duration: 3.0Hrs **Minor Test I : 20**
Minor Test II : 20
Major Test : 60

Course Outcomes: Human Molecular Genetics is a vast field that provides information of Genetic Material, general principles and applications of molecular hybridization. It provides comprehensive guide to the structure, function and changes in the human genome.

CO-1: Introduction to the structure of Gene and Genome
CO-2: Structure and functions of Nucleic Acids
CO-3: Concept of transcription and translation
CO-4: Introduction to Mutations and its types.
CO-5: Understanding the genome evolution and various genome projects.

Unit-I **(13hrs)**

Introduction to Gene and Chromosomes

1.1Introduction to chromosomes
- 1.1.1 Structure and types of Chromosomes
- 1.1.2 Nucleosome, solenoid, chromosomal territories
- 1.1.3 Special Chromosomes: Polytene chromosomes**,** satellite chromosomes and Lampbrush chromosomes

1.2 Chromatin: Structure, Functional states of chromatin
1.3 Histone Modifications: Methylation (cpG islands) and Acetylation
1.4 Holocentric chromosome**,** Heterochromatin and Euchromatin
1.5 An overview of Genome

Unit-II **(13hrs)**

Nucleic Acid: Structure & functions

2.1 DNA-Structure & types
2.2 RNA- Structure and types
2.3 Difference between DNA and RNA
2.4 DNA as Genetic Material
2.5 DNA Replication in Prokaryotes & Eukaryotes
2.6 Mechanism of Transcription and its factors
2.7 Post-transcriptional modification:
- 2.7.1Capping
- 2.7.2 Polyadenylation
- 2.7.3 Splicing
- 2.7.4 RNA editing

Unit-III **(12hrs)**

Gene Regulation

3.1 Mechanism of Translation (Initiation, Elongation & Termination)
3.2 Post translation modifications
3.3 Positive and Negative Regulation (Enhancers and Junk DNA)
3.4 Gene Regulation in Eukaryotes
- 3.4.1 Transcriptional gene regulation
- 3.4.2 Post-transcriptional gene regulation
- 3.4.3 Translational gene regulation
- 3.4.4 Post-translational gene regulation

M.Sc. Human Genetics

I[st] Semester (CBCS)

Syllabus for the examination to be held in December, 2022, 2023, 2024

Course No. PSHGTC-108 **Course Title: Human Molecular Biology**
Credits: 04 **Maximum Marks: 100**
Time Duration: 3.0Hrs **Minor Test I : 20**
Minor Test II : 20
Major Test : 60

Unit-IV

Mutations **(12hrs)**

4.1 Mutations

4.1.1 Types of mutations; Base Substitutions, deletions and insertions, Non sense, missense, silent, Point, Frameshift Mutations,Germ line and somatic Mutations.

4.1.2 Physical, Chemical and Biological Mutagens

4.2 DNA repair mechanism

4.2.1 Direct Repair

4.2.2 Excision Repair, Base Excision Repair, Nucleotide Excision Repair, Mismatch Repair, Non homologous end Joining, SOS repair

Unit-V **(13hrs)**

Human Genome

1.1 Basic concept of Human Genome

1.1.1 History, Organization and goals of Human Genome

1.1.2 Human genome diversity and its importance

1.1.3 Human Genome Project

1.2 Homology, Paralogs & Orthologs

1.3 Repetitive DNA and its types

1.4 Transposable elements

1.5 Genome evolution

1.5.1 Nuclear genome evolution

1.5.2 Mitochondrial genome evolution

1.5.3 Sex chromosome evolution

Note for paper setting:

Examination theory weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1.5 Hrs	20
Minor Test-II	21-40%	1.5 Hrs	20
Major Test	41-100%	3.0 Hrs	60

i. Major test will have two sections (A&B)

ii. Section A is compulsory comprising of 10 questions of 1.5 marks each and be spread over entire syllabus

iii. Section B comprises of 6 questions (2 from each unit) from the remaining 3 units and candidate has to attempt one question from each unit (15 marks each).

BOOKS RECOMMENDED

1) Friedberg et al, DNA repair & Mutagenesis, 2006
2) Benjawin Lewin, Gene IX. Jones and Barlett Publishers. 2008
3) F Vogel A.G. Motulusky Human Genetics: Problems and Approaches. 5th Edition, BMC, 2010
4) T. A. Brown,: Gene Cloning: 7th Edition, Garland Science, 2010
5) Tom Strachen, Human Molecular Genetics, 4th Edition, Garland Science, 2010.
6) D. Peter Snustad and Michael J. Simmons. Principles of Genetics. 6th edition. John Wiley & Sons, Inc.,2011.
7) Robert J Brooker, Genetics- Analysis and Principles, 2012
8) T. A. Brown,: Gene Cloning: 7th Edition, Garland Science, 2013
9) Arumugam *et al*, Molecular biology & Genetic Engineering, 2014
10) Thompson & Thompson Genetics in Medicine; 8th edition, Imprint : Saunders, 2015.
11) Lewin, Gene XII, 12th Edition, 2017.

M.Sc. Human Genetics

I[st] Semester (CBCS)

Syllabus for the examination to be held in December, 2022, 2023, 2024

Course No. PSHGTC-109 **Course Title: - Principles of Genetics**
Credits: 02 **Maximum Marks : 50**
Time Duration: 2.5Hrs **Minor Test I : 05**
Minor Test II : 05
Major Test : 40

Course Outcome: The course has been designed to provide an introduction to the basic concepts of Genetics. The course will help the students to understand the mechanism of determining sex of an individual. Structural details and the role of chromosomes in human congenital anomalies have been discussed so that a student pursuing P.G course in Human Genetics is able to understand the importance of human chromosomes.
CO-1: Understanding about history of cytogenetics and extensions of Mendelism.
CO-2: Introduction to the Structure of Human Chromosomes and related conditions.
CO-3: Knowledge about chromosomal theory of inheritance.

Unit-I
Hereditary and Variations **(10 hrs)**

1.1 Cytogenetic: Introduction and its importance.
1.2 Mendel's Laws of Heredity: Law of segregation & Law of Independent Assortment
1.3 Deviation of Mendelian laws
1.3.1 Incomplete Dominance
1.3.2 Co-dominance
1.3.3 Multiple allelism, Lethal alleles
1.3.4 Penetrance and expressivity
1.3.5 Epistasis and its types
1.3.6 Pleiotropy
1.4 Chromosomal Theory of Heredity, Inheritance and Non disjunction as a proof to Chromosomal Theory

Unit-II **(10hrs)**
Chromosomal Nomenclature and Sex determination

2.1Nomenclature of Chromosomes
2.2 Landmarks of Chromosomes
2.3 Sex determination & differentiation in Humans
2.4 Inactivation of X chromosomes
2.5 Dosage compensation- Lyon Hypothesis
2.6 Non chromosomal basis of sex determination

Unit -III **(10hrs)**
Genetic Inheritance

3.1 Linkage and crossing over Introduction
3.1.1 Genetic mapping and LOD score analysis
3.2 Interference and Coincidence
3.3 Quantitative Inheritance
3.4 Extra Nuclear inheritance
3.5 Sex-limited, sex linked and sex influenced traits.
3.6 Pedigree analysis
3.6.1 Symbols, Construction and patterns of Inheritance
3.6.2 Complications to the basic pedigree patterns (Non-Penetrance, Variable Expressivity, Pleiotrophy etc)

M.Sc. Human Genetics

I[st] Semester (CBCS)

Syllabus for the examination to be held in December, 2022, 2023, 2024

Course No. PSHGTC109 **Course Title: - Principles of Genetics**
Credits: 02 **Maximum Marks : 50**
Time Duration: 2.5Hrs **Minor Test I : 05**
Minor Test II : 05
Major Test : 40

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	5
Minor Test-II	21-40%	1 Hr	5
Major Test	41-100%	2.5 Hrs	40

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

Books Recommended:

1. F Vogel A.G. Motulusky. Human Genetics: Problems and Approaches. 5th Edition, BMC, 2010.
2. Ricky Lewis, Concepts of Human Genetics, 2011.
3. Bruce R. Kork and Mira B Irons, Human Genetics & Genomics (4th edition), 2013.
4. ABC of Clinical genetics, Helen M Kingston, 4th Edition, BMJ, 2015.
5. Robert Nussbaum et al. Thompson & Thompson genetics in Medicine, 8th Edition, Elsevier, 2015.
6. Robert L. Nussbaum, Roderick R. Mcinnes, & Huntington F. Willard, Thompson & Thompson Genetics in Medicine;, 8th edition, Imprint : Saunders, 2015.
7. Human Heredity : Principles and Issues by Micheal R. Cummings; 11th edition, Cengage Learning,2016.
8. Emerys & Rimoin, Principles & Practice of Medical Genetics, 7th Edition, Elsevier, 2018

M.Sc. Human Genetics

I[st] Semester (CBCS)

Syllabus for the examination to be held in December, 2022, 2023, 2024

Course No. PSHGTC-110 **Course Title: - Gene diagnostics and Methodology**

Credits: 02 **Maximum Marks : 50**

Time Duration: 2.5 Hrs **Minor Test-I : 05**

Minor Test-II : 05

Major Test : 40

Course Outcomes: The course has been designed to provide an introduction to the basic concepts about the different types of diagnostic techniques. These techniques are the most relevant techniques used for the genetic testing purposes and would give a more comprehensive picture to make genetic testing relevant in day to day life.

CO-1: Introduction to basic research methodologies - Centrifugation, Electrophoresis and microscopy.

CO-2: Knowledge about various cytogenetic techniques.

CO-3: Understanding various types of molecular diagnostic techniques.

Unit-I **(10hrs)**

Research Methodology

1.1 Centrifugation: Basic principle, Types (Simple & Ultracentrifuge; types of rotors) and its Applications

1.2 Electrophoresis: Principle, Types and Applications

1.3 Spectrophotometer: Principle, Working and Applications

1.4 Microscopy: Principle, Working, Application of light, electron, Phase contrast and fluorescence microscope, SEM and TEM.

1.5 Blotting techniques: Northern, Southern and Western Blotting and Dot Blot assay.

1.6 Chromatography techniques: Paper chromatography, Liquid chromatography, Gas chromatography, Thin Layer Chromatography (TLC)

Unit-II **(10hrs)**

Cytogenetic Techniques

2.1 Overview of Cytogenetic Techniques

2.2 Lymphocyte Culturing

2.3 Chromosome Banding Techniques: G banding, Q banding, R banding, C banding, NOR banding, High Resolution banding

2.4 Conventional Karyotyping

2.5 Molecular Cytogenetic Techniques

- 2.5.1 Conventional FISH
- 2.5.2 Spectral Karyotyping (SKY)
- 2.5.3 MLPA
- 2.5.4 Comparative Genomic Hybridization (CGH)
- 2.5.5 Genomic In Situ Hybridization (GISH)

Unit-III **(10 hrs)**

Molecular diagnostic techniques.

3.1 Common Molecular Techniques

- 3.1.1 PCR: Types (Real time-PCR, Multiplex PCR, reverse transcriptase-PCR, Nested PCR, RFLP, AFLP and Isothermal PCR) and applications
- 3.1.2 DNA Fingerprinting
- 3.1.3 Lateral flow methods

3.2 DNA Sequencing

- 3.2.1Maxam-Gilbert (Chemical)
- 3.2.2 Sanger sequencing method
- 3.2.3 Massive parallel sequencing (NGS and pyrosequencing)

M.Sc. Human Genetics

Ist Semester (CBCS)

Syllabus for the examination to be held in December, 2022, 2023, 2024

Course No. PSHGTC-110 **Course Title: - Gene diagnostics and Methodology**

Credits: 02 **Maximum Marks : 50**

Time Duration: 2.5Hrs **Minor Test-I : 05**

Minor Test-II : 05

Major Test : 40

3.2.4 SNPs (rsIDS) and SNP databases.

3.3 Genetic diagnosis through screening

3.3.1 Population Screening: Methods and types

3.3.2 Family segregation

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	5
Minor Test-II	21-40%	1 Hr	5
Major Test	41-100%	2.5Hrs	40

I. Major test will have two sections (A&B)

II. Section A is compulsory comprising of 10 questions of 1 mark each and be spread over entire syllabus

III. Section B comprises of 4 questions from the remaining 2 units and candidate has to attempt one questionfrom each unit (10 marks each).

Books Recommended:

1. David L Speeta and Bobert D, Basic Method in Microscopy, 2006.
2. Cox & Sinclair, Molecular Biology in Medicine, Blackwell,. 2009.
3. DeGrouchy & Turleau, Clinical Atlas on Human Chromosomes, Wiley, 2010.
4. Jankowski & Polak, Clinical Gene Analysis and Manipulation , Cambridge, 2011.
5. Korf, Human Genetics- A Problem Based Approach, Blackwell, 2011.
6. Ricki Lewis. Human Genetics- Concepts and Application, 11th edition. WCB- McGraw Hill, 2011.
7. Andreas Hofmann and Samuel Clokie, Wilson and Walker's Principles and Techniques of Biochemistry and Molecular Biology, Cambridge University Press Edition 8th, 2018.

Ist Semester (CBCS)

M.Sc. Human Genetics
Syllabus for the examination to be held in December, 2022, 2023, 2024

LAB COURSE NO. PSHGPC-106

List of practicals based on Theory Course no. 101 & 102.

1. To study the different parts of the light microscope.
2. To study the working and principle of light microscope.
3. Determination of bleeding and clotting time.
4. To determine the blood groups & Rh factor of your own blood.
5. To study different stages of Mitosis & Meiosis.
6. To study the T.S. of Human Pancreas.
7. To study the T.S. of Human Thyroid follicles.
8. To study the T.S. of Human Ovary.
9. To study the T.S. of Human Sperm.
10. To study the T.S. of Adrenal gland.
11. To study the T.S. of Testis.
12. Identify and study the different types of Bones- Long & Short bones.
13. To study the different types of girdles-Pectoral & Pelvic.
14. To study the different bones of Human Skull.
15. To study the different types of joints in Humans.
16. To measure blood pressure by Sphygmomanometer.
17. To measure Mean arterial pressure and pulse pressure estimation.

LAB COURSE NO. PSHGPC-107

List of practicals based on Theory Course no. 108, 109 & 110

1. To study the different biosafety levels and good lab practices.
2. To perform Sterilization of glassware and plasticware.
3. To study the study the presence of Drumstick in human neutrophils cells to understand the process of X chromosome inactivation.
4. To study the study the presence of Barr body in human buccal epithelial cells to understand the process of X chromosome inactivation.
5. To study Mendel's Law of Hereditary and its exception.
6. To study the procedure for Human Lymphocyte culturing from whole blood.
7. To scan the provided slides for a well spread metaphase plate in order to identify different types of human chromosomes.
8. Identify and comment on the provided photographs of the suspected patients
   a. Down syndrome
   b. Edward syndrome
   c. Patau syndrome
   d. Turner syndrome
   e. Klinefelter syndrome
9. To study the scheme of the Karyotype preparation
10. Preparation of Karyotype of Normal male from the provided photographs of metaphase plates.
11. Preparation of Karyotypes of Normal female from the provided photographs of metaphase plates.
12. Identify and study the different types of equipments required for DNA isolation.
13. Preparation of the chemicals required for DNA isolation.

14) Preparation of stock solutions for DNA isolation.

15) Preparation of working solutions from stock solutions for DNA isolation.

16) To carry out the DNA extraction from the saliva sample.

17) Preparation of Agarose gel for the Electrophoresis.

18) To carry out the Quantitative analysis of the isolated DNA via Gel Electrophoresis.

19) To carry out the Quantitative analysis of the isolated DNA via Spectrophotometer.

20) To study the principle and working of centrifugation.

21) To study principle and procedure of ELISA.

22) Demonstration of Thermocycler and RT-PCR.

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2023, 2024, 2025

Course No. PSHGTC- 201 **Course Title: Biochemistry of Metabolic Disorders & Developmental Genetics**

Credits: 04 **Maximum Marks : 100**

Time Duration: 3.0Hrs **Minor Test I : 20**

Minor Test II : 20

Major Test : 60

Course Outcomes (CO)

CO-1: The syllabus has been designed to provide the students about the carbohydrate metabolism pathways inhuman body.

CO-2: The students will get knowledge about Protein and Nucleic acid metabolism. Any change in biochemicalpathways leads to the change in the product and the same gets reflected in the form of change in the phenotype**.**

CO-3: Students will be able to understand the metabolism of Lipids and fatty acid and their related disorders.

CO-4: The course will also help students to understand the process of development at genetic level.

CO-5: The course offers understanding about differentiation of human reproductive systems and focuses on geneticbasis of reproductive disorders.

Unit–I

1.1 Introduction to Carbohydrate Metabolism **(12 hrs.)**

1.2 Disorders of Carbohydrate Metabolism (Genetic cause, diagnosis & treatment):

- 1.2.1 Lactose Intolerance
- 1.2.2 Glucose-6 Phosphate dehydogenase deficiency (G-6PDD)
- 1.2.3 Fructose Intolerance
- 1.2.4 Diabetes Mellitus
- 1.2.5 Galactosemia

Unit–II

2.1 Introduction to Proteins & Amino acids. **(13 hrs.)**

2.2 Disorders of Amino acids metabolism (Genetic cause, diagnosis & treatment):

- 2.2.1 Phenylketonuria
- 2.2.2 Alkaptonuria
- 2.2.3 Tyrosinemia
- 2.2.4 Albinism

2.3 Metabolic Disorders of Purines and Pyrimidines (Genetic cause, diagnosis & treatment):

- 2.3.1 Hyperuricemia
- 2.3.2 Lesch-Nyhan Syndrome

2.4 Metabolic disorders of Porphyrin (Genetic cause, diagnosis & treatment):

- 2.4.1 Acute Intermittent Porphyrin
- 2.4.2 Erythropoietic Porphyria

2.5 Metabolic disorders of Glycosamineglycans & Glycoproteins (Genetic cause, diagnosis & treatment):

- 2.5.1 Mucopolysaccharidosis
- 2.5.2 Mucolipidosis

Unit–III

3.1 Introduction to Lipids, & Fatty acids and their metabolism **(13 hrs.)**

3.2 Disorders of Lipid Storage (Genetic cause, diagnosis & treatment):

- 3.2.1 Tay Sachs Disease
- 3.2.2 Krabbe Disease

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2023, 2024, 2025

Course No. PSHGTC- 201 **Course Title: Biochemistry of Metabolic Disorders & Developmental Genetics**

Credits: 04 **Maximum Marks : 100**

Time Duration: 3.0 Hrs **Minor Test I : 20**

Minor Test II : 20

Major Test : 60

3.3 Disorders of Fatty acid Metabolism (Genetic cause, diagnosis & treatment):

3.3.1 Hyperlipidemia

3.3.2 Hypercholesterolemia

Unit-IV **(12 hrs)**

4.1 Introduction to development gene family disorders.

4.2 Limb as a development model.

4.3 Role of Development genes in cancer.

4.4 Developmental malformations.

4.5 Hydatiform moles.

4.5.1 Complete molar pregnancy

4.5.2 Partial molar pregnancy

4.6 Teratogen induced reproductive complications.

4.7 Gene-teratogens interactions.

Unit-V

5.1. Male and Female reproductive system **(13 hrs)**

5.1.1. Gonads and differentiation of reproductive systems

5.1.2. Hormonal regulation of sexual differentiation

5.2. Reproductive disorders

5.2.1. Pseudohermaphroditism

5.2.2. True hermaphroditism

5.2.3 Gonadal dysgenesis

5.2.4. Anomalies of genital ducts

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1.5Hrs	20
Minor Test-II	21-40%	1.5Hrs	20
Major Test	41-100%	3.0 Hrs.	60

i. Major test will have two sections (A&B)

ii. Section A is compulsory comprising of 10 questions of 1.5 marks each and be spread over entire syllabus

iii. Section B comprises of 6 questions (2 from each unit) from the remaining 3 units and candidate has to attemptone question from each unit (15 marks each).

BOOKS RECOMMENDED

1. T. Subramanium Molecular Developmental biology, 2008.
2. Mathews et al.: Biochemistry (4rd Ed.), Pearson, 2012.
3. Harpers illustrated Biochemistry (31st edition) The Mc Graw Hill Companies, 2014.
4. Berg et al.: Biochemistry (8th Ed.), Freeman, 2015.
5. Biochemistry by Donald Voet (5th edition) pubisher : Wiley, 2016.
6. Lubert Stryer's; Biochemistry, 8th Edition, published by W.H. Freeman and Company, 2016.
7. Lehninger Principles of Biochemistry (7th Ed.), MacMillan Worth, 2017.
8. Harpers illustrated Biochemistry (31st edition) The Mc Graw Hill Companies, 2018.
9. Harpers illustrated Biochemistry (31st edition) The Mc Graw Hill Companies, 2018

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2022, 2023 and 2024

Course No. PSHGTC- 207 **Course Title: - Microbial Genetics and Clinical Biology**

Credits: 02 **Maximum Marks : 50**

Time Duration: 2.5Hrs **Minor Test I : 05**

Minor Test II : 05

Major Test : 40

Course Outcomes (CO)

CO-1: Introduction to Microbiology.

CO-2: Methods of gene transfer and role of vectors with their property.

CO-3: This course is about various pathogens like bacteria, virus and fungi and about their testing

Unit-1

Introduction to Microbiology **(10 hrs)**

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: size, shape, and arrangement of bacteria.
1.4 Morphology and ultra structure of Virus: size, shape, and arrangement of Virus.
1.5 Morphology and ultra structure of Fungus: size, shape, and arrangement of Fungus.
1.6 Microbial nutrition and growth curves.
1.7 Toxins: Exotoxins and Endotoxins

Unit- II **(10 hrs)**

2.1 Methods of genetic transfers – transformation, conjugation.
2.2 Transduction: generalized and specialized transduction.
2.2.1 Phage conversion.
2.2.2 Sexduction (F -duction)
2.2.3 Mapping genes by interrupted mating.
2.3 Plasmid vectors and their properties
2.3.1 Expression cloning, Cloning vectors, BACs, YACs
2.4 Plasmid: types and their significance
2.5 EMSA and Luciferase assay

Unit-III **(10hrs)**

Introduction to Pathogens

3.1 Microbial diseases
3.1.1 Bacterial Disease: TB, Cholera, Pneumonia, Diarrhorea
3.1.2 Viral diseases: AIDS, Hepatitis, COVID-19
3.1.3 Fungal disease: Candidiasis, Aspergillosis, blastomycosis
3.2 Microbial markers
3.3 Microbiome
3.3.1Micro biome –Introduction
3.4 Gut micro biome and its function.
3.5 Microbiome and Human Disease
3.6 Microbiome testing

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2022, 2023 and 2024

Course No. PSHGTC- 207 **Course Title: - Microbial Genetics and Clinical Biology**

Credits: 02 **Maximum Marks : 50**

Time Duration: 2.5Hrs **Minor Test I : 05**

Minor Test II : 05

Major Test : 40

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1.0 Hrs	5
Minor Test-II	21-40%	1.0 Hrs	5
Major Test	41-100%	2.5Hrs	40

I. Major test will have two sections (A&B)

II. Section A is compulsory comprising of 10 questions of 1 mark each and be spread over entire syllabus

III. Section B comprises of 4 questions from the remaining 2 units and candidate has to attempt one questionfrom each unit (10 marks each).

BOOKS RECOMMENDED

1. Richard Coico, Geoffrey Sunshine, Immunology (A short course), 6th Edition, 2008.
2. Kenneth Murphy et al., Immunobiology, 8th GS publications, 2012.
3. Immunology by Richard A. Goldsby (Editor), Barbara A. Osborne, Thomas J. Kindt, Janis Kuby, JanisKuby, Richard A. Goldby, 7th edition, 2013.
4. Prescott, Harley, Klein; Microbiology, 10th edition, Mc Graw- Hill Higher Education, 2017.
5. Pelczar, Michael J. Jr. / Chan, E.C.S / Krieg, Noel R., Microbiology, 5th Edition , Mc Graw- Hill HigherEducation, 2017.
6. Roitt's, Essential Immunology, 13th edition, Wiley-Blackwell Co., 2017.
7. Robert Rich et al. Clinical Immunology, Elsevier, 5th Edition, 2018.
8. Kenneth Murphy et al., Janeways Immunobiology, 9th GS publications, 2019.

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2022, 2023 and 2024

Course No. PSHGTC- 208 **Course Title: Immunogenetics**
Credits: 04 **Maximum Marks: 100**
Time Duration: 3.0 Hrs **Minor Test I : 20**
Minor Test II : 20
Major Test : 60

Course outcome (CO)

CO-1: - Objectives of this course is to understand basic principles of Immunology
CO-2: The course will help the students to gather knowledge about Humoral and cell mediated Immunity
CO-3: The course is about the Immunological disorders.
CO-4: The objective of the course is apprise students about the basics of Immunodiagnostic techniques
CO-5: Course has been designed to make the student of Human Genetics Familiar with Immunobiotechnology which discusses its applications.

Unit – I **(13hrs)**

Introduction to the Immune System

1.1 Introduction to immune system, Innate and acquired immunity.
1.2 Clonal nature of immune response.
1.3 Organization and structure of lymphoid organs
1.4 Cells of the immune system: Hematopoiesis and differentiation, B- lymphocytes, T -lymphocytes.
1.5 Macrophages, Dentritic cells, Natural killer and Lymphokine activated killer cells, Eosinophils, Neutrophils and Mast cells.
1.6 Nature and Biology of antigens and super antigens.
1.7 Antibody structure and function, antibody mediated effector functions, antibody classes and biological activity
1.8 Antigenic determinants on immunoglobulins, Immunoglobulin superfamily, BCR & TCR, generation of antibody diversity.

UNIT – II **(12hrs)**

Humoral and cell mediated immunity

2.1 Regulation of immune response
2.2 Antigen processing and presentation, generation of humoral and cell mediated immune responses
2.3 Activation of B- and T- lymphocytes
2.4 Complement System: components of complement, complement activation, complement cascade, regulation of Complement System
2.5 Cytokines, cytokines receptors, cytokines antagonists, role of cytokines in T_H1/T_H2 subset development and their role in immune regulation.
2.6 MHC: MHC molecules and genes, MHC restriction,
2.7 Cell-mediated cytotoxicity: Mechanism of T cell and NK cell mediated lysis.
2.8 Antibody dependent cell mediated cytotoxicity, macrophage mediated cytotoxicity.

Unit- III **(13hrs)**

Immunological disorders

3.1 Autoimmunity and auto immune disorders, immunological tolerence.
3.2 Organ specific and systemic autoimmune diseases, animal models for autoimmune diseases and the molecular mechanism, immunodeficiency disorder- AIDS
3.3 Hypersensitivity: IgE mediated Hypersensitivity, Antibody mediated cytotoxic Hypersensitivity, Immune complex- mediated Hypersensitivity, Delayed type Hypersensitivity

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2022, 2023 and 2024

Course No. PSHGTC- 208 **Course Title: Immunogenetics**
Credits: 04 **Maximum Marks: 100**
Time Duration: 3.0Hrs **Minor Test I : 20**
Minor Test II : 20
Major Test : 60

3.4 Transplantation immunology: Immunological basis of graft rejection, clinical manifestation of graft rejection, general Immunosuppressive therapy, specific immunosuppressive therapy, immune tolerance to allografts.
3.5 Immunological tolerance; central tolerance, peripheral tolerance, component of peripheral tolerance

Unit - IV **(12hrs)**
Immunodiagnostic procedures
4.1 Antibody interactions and Techniques
4.1.1 ELISA and its variants
4.1.2 ELISPOT
4.1.3 Radio immunoassay
4.1.4 Immunofluorescence.
4.2 Flow cytometry and Fluorescence, Immuno electron microscopy
4.3 Agglutination and haemagglutination assays
4.4 Types of immundiffusion and immuno electrophoretic procedures, isolectric focusing
4.5 Affinity chromatographic methods and Immunoblotting.

Unit-V **(13hrs)**
Immunobiotechnology
5.1 Hybridoma Technology and Monoclonal antibodies detection and application of monoclonal antibodies;
5.2 Lymphokines: production and applications,
5.3 Interleukine therapy
5.4 Vaccines
5.4.1 History of vaccine development
5.4.2 Introduction to the concept of vaccine.
5.5 Active and passive immunization,
5.6 Designing vaccines for active immunization:
5.7 Conventional vaccines, subunit vaccines, conjugate vaccines, DNA vaccines.
5.8 Recombinant vector vaccines
5.9 Cell culture and maintenance of cell lines

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in Examination	Time allotted	% marks
Minor Test-I	Upto 20%	1.5Hrs	20
Minor Test-II	21-40%	1.5Hrs	20
Major Test	41-100%	3.0 Hrs	60

i. Major test will have two sections (A&B)
ii. Section A is compulsory comprising of 10 questions of 1.5 marks each and be spread over entire syllabus
iii. Section B comprises of 6 questions (2 from each unit) from the remaining 3 units and candidate has to attempt one question from each unit (15 marks each).

BOOKS RECOMMENDED

1) Goldsby, R. A., Kindt, T.J. and Osborne,B.A. (2002).Kuby Immunology. W.H. Freeman and company, New York.
2. Coleman, R.M., Lombard, M.F. and Sicard, R.E.(1992).Fundamental Immunology. Wm.C.Brown publishers,USA.
3. Roitt,I., Brostoff, J. and Male,D. (1999). Immunology. Hartcourt Brace and Company , Asia Pte.Ltd.
4. Benjamini,E.,Coico,R., and Sunshine, G. (2000). Immunology – a short course. John Wiley and Sons. Inc., New York.
5. Davies,H. (1997). Introductory Immunology. Chapman and Hall, New York
6. Bratke & Myrtek (2007). Immunology : The experimenter series.Elsener Pub.
7. Wood, Peter (2008). Understanding Immunology Elseiver Pub. 2 nd edition

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2022, 2023 and 2024

Course No. PSHGTC-209 **Course Title: - Human Molecular Genetics**

Credits: 04 **Maximum Marks : 100**

Time Duration: 3.0 Hrs **Minor Test I : 20**

Minor Test II : 20

Major Test : 60

Course Outcomes

CO-1: The course has been designed with the objective to make the students of Human Genetics to learn various hybridized assays and cloning and its applications on human diseases.

CO-2: Students will get knowledge about Molecular pathology of human diseases.

CO-3: The course will provide knowledge about Identification of diseases and instability of genome.

CO-4: Students will be able to learn various advanced techniques for detection of proteins and gene analysis

CO-5: The students will learn various diagnostic and therapeutic approaches for human diseases

Unit-I **(12hrs)**

1. Molecular Hybridization

1.1 DNA hybridization assays

1.2 Nucleic acid probe

1.3 Principles of molecular hybridization

1.3.1 Methods and applications of molecular hybridization.

1.3.2 Synthesis and labelling of probes.

1.4 Enzymes used in RDT: Restriction Endonuclease, Other enzymes

1.5 Gene cloning

1.5.1 Ethical issues in gene cloning

1.6 Applications of RDT in Human Diseases

Unit-II

Molecular Pathology **(13 hrs)**

2.1. Introduction.

2.2. Rules for nomenclature of mutations and databases of mutation.

2.3. Loss and gain of function mutations.

2.4. Molecular pathology: from gene to disease.

2.5. Molecular pathology: from disease to gene.

2.6. Molecular pathology of chromosomal disorders.

2.7. Epigenetics and its role in Human diseases.

UNIT –III **(12hrs)**

Identification of disease associated genes

3.1 Principles and strategies for identifying human disease genes: 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.

3.5 Pathogenicity associated with repeated sequence

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2022, 2023 and 2024

Course No. PSHGTC-209 **Course Title: - Human Molecular Genetics**

Credits: 04 **Maximum Marks : 100**

Time Duration: 3.0 Hrs **Minor Test I : 20**

Minor Test II : 20

Major Test : 60

UNIT-IV

Genome Sequence and Function **(13hrs)**

4.1 Human Genome Project

4.1.1 1000 Genome Project

4.1.2 100,000 Genome Project

4.1.3 Pangenome Project

4.2 Studying the Transcriptome.

4.2.1 Studying Transcriptome by Microarray.

4.2.2 Studying Transcriptome by Sequence Analysis.

4.2.3 Introduction of NGS.

4.3 Studying the Proteome

4.4 Protein Profiling (2D Electrophoresis & MALDI TOF)

4.5 Protein degradation

Unit- V **(13hrs)**

Diagnostics and Therapeutics

5.1 Gene therapy

5.1.1 Concept of Gene therapy (ex-vivo and in- vivo approach)

5.1.2 Vectors used in gene therapy

5.1.3. Biological vectors- retrovirus, adenovirus, herpes

5.2 Gene Editing (CRISPR)

5.2.1 Genome applications

5.2.2 Genome editing treatments

5.3 Pharmacogenomics

5.3.1 Concepts of Pharmacogenomics.

5.3.2 Pharmacogenomics in drug discovery and drug development.

5.4 Enzyme therapy and its applications.

5.5 Animal models for the study of human genetic diseases: Drosophila, Yeast, Mouse, Neurospora.

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1.5Hrs	20
Minor Test-II	21-40%	1.5Hrs	20
Major Test	41-100%	3.0 Hrs.	60

i. Major test will have two sections (A&B)

ii. Section A is compulsory comprising of 10 questions of 1.5 marks each and be spread over entire syllabus

iii. Section B comprises of 6 questions (2 from each unit) from the remaining 3 units and candidate has toattempt one question from each unit (15 marks each).

Books Recommended:

1. F Vogel A.G. Motulusky. Human Genetics: Problems and Approaches. 5th Edition, BMC, 2010.
2. ABC of Clinical genetics, Helen M Kingston, 4th Edition, BMJ, 2015.
3. Robert L. Nussbaum, Roderick R. McInnes, & Huntington F. Willard, Thompson & Thompson Genetics inMedicine;, 8th edition, Imprint : Saunders, 2015.
4. Human Heredity : Principles and Issues by Micheal R. Cummings; 11th edition, Cengage Learning, 2016.
5. Emerys & Rimoin, Principles & Practice of Medical Genetics, 7th Edition, Elsevier, 2017.
6. R.J. Mc Kinlay Gardner, Grant R.Sutherland Chromosome Abnormalities and Genetic Counselling (Oxford Monographs on Medical Genetics), 5th edition,. Oxford University Press, USA. 2018.

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2022, 2023 and 2024

Course No. PSHGTC-210	**Course Title: Advanced Cytogenetics**
Credits: 02	**Maximum Marks : 50**
Time Duration: 2.5Hrs	**Minor Test I : 05**
	Minor Test II : 05
	Major Test : 40

Course Outcomes

CO-1: The course has been designed with the objective to make the students of Human Genetics to learn about Structural and numerical abnormality.

CO-2: Students will get knowledge about various human congenital abnormalities

CO-3: The course will provide knowledge about mutagens and teratogens

.

Unit – I **(10hrs)**

Chromosomal Anomalies-I

1.1 Introduction to chromosomal Anomalies: Structural and Numerical

1.2 Structural Alterations in chromosomes: Deletions, Duplications, Translocations, Inversions.

1.3 Numerical Alterations in chromosomes: Aneuploidy, Polyploidy

1.4 Numerical Abnormalities of Autosomes Down Syndrome, Edward Syndrome & Patau Syndrome

1.5 Numerical Abnormalities of Sex Chromosomes: Turner syndrome & Klinefelter syndrome.

1.6 Human Artificial chromosomes

Unit – II **(10hrs)**

Human Congenital Abnormalities

2.1 Introduction.

2.2 Neural Tube Defects.

- 2.2.1Anencephaly
- 2.2.2 Encephalopathy
- 2.2.3 Hydranencephaly

2.3 Spina bifida including myelomeningocele and others

2.4 Cleft Lip/Cleft Palate.

2.5 Genomic Imprinting/ Uniparental Disomy

- 2.5.1 Prader-Willi Syndrome
- 2.5.2 Angelman Syndrome
- 2.5.3 Beckman Weidworth Syndrome

Unit-III **(10hrs)**

3.1Chromosome Instability syndrome

- 3.1.1Ataxia Telangiectasia,
- 3.1.2Fanconi Anaemia
- 3.1.3Bloom's Syndrome
- 3.1.4 Nijmegen breakage syndrome

3.2 Effect of mutagenic and Teratogenic exposures in early pregnancy.

3.3 Effects of chromosomal instability on human health.

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2022, 2023 and 2024

Course No. PSHGTC-210 **Course Title: Advanced Cytogenetics**

Credits: 02 **Maximum Marks : 50**

Time Duration: 2.5Hrs **Minor Test I : 05**

Minor Test II : 05

Major Test : 40

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hrs	05
Minor Test-II	21-40%	1Hrs	05
Major Test	41-100%	2.5Hrs	40

I. Major test will have two sections (A&B)

II. Section A is compulsory comprising of 10 questions of 1 mark each and be spread over entire syllabus

III. Section B comprises of 4 questions from the remaining 2 units and candidate has to attempt one questionfrom each unit (10 marks each).

Books Recommended:

1. F Vogel A.G. Motulusky. Human Genetics: Problems and Approaches. 5th Edition, BMC, 2010.
2. ABC of Clinical genetics, Helen M Kingston, 4th Edition, BMJ, 2015.
3. Robert L. Nussbaum, Roderick R. Mcinnes, & Huntington F. Willard, Thompson & Thompson Genetics inMedicine;, 8th edition, Imprint : Saunders, 2015.
4. Human Heredity : Principles and Issues by Micheal R. Cummings; 11th edition, Cengage Learning, 2016.
5. Emerys & Rimoin, Principles & Practice of Medical Genetics, 7th Edition, Elsevier, 2017.
6. R.J. McKinlay Gardner, Grant R.Sutherland Chromosome Abnormalities and Genetic Counselling (Oxford Monographs on Medical Genetics), 5th edition,. Oxford University Press, USA. 2018.

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2022, 2023 and 2024

Credits: 04 **Max.Marks:100**

LAB COURSE NO. PSHGPC-205

List of practicals based on Theory Course no. 201 , 207, 208

1. To study the principle, construction and uses of Laminar air flow.
2. To study the principle, construction and uses of autoclave.
3. To study the principle, construction and uses of hot air oven.
4. To study the working and principle of auto-analyzer:
    i. Semi-auto analyzer
    ii. Fully auto analyzer
5. Qualitative and quantitative estimation of carbohydrates, Lipids and proteins.
6. Plasma and serum isolation.
7. To perform liver function test of a given serum/blood sample on auto analyzer.
8. To perform lipid profiling of a given serum/blood sample on auto analyzer.
9. To perform renal profiling of a given serum/blood sample on auto analyzer.
10. To study different types of cells from blood smear.
11. To study the common apparatus used in microbiology.
12. To study the general morphology of bacteria.
13. To prepare chemicals for bacterial staining.
14. To prepare a temporary mount of bacteria (lactobacillus) present in curd.
15. To prepare the different culture media.
16. To carry out various biochemical tests for Lactobacillus bacteria-
    i. Gram Staining
    ii. Oxidase Test
    iii. Catalase Test
17. To observe the presence of any bacteria belonging to Enterobacteriaceae family-
    i. IMViC Test-
    i. Indole Test
    ii. Methyl Test
    iii. Voges-Proskauer Test
    iv. Citrate Test
18. To carry out the extraction of bacterial DNA
19. To carry out the amplification of bacterial DNA by Polymerase Chain Reaction
20. To study the abnormal conditions of genital duct (Hypospadiasis, Anorectal malformations, Ambiguousgenitalia).

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May 2022, 2023 and 2024

2nd Semester (CBCS)

Credits: 04 **Max.Marks:100**

LAB COURSE NO. PSHGPC-206

List of practicals based on Theory Course no. 209 & 210

1. To prepare chemicals required for GTG banding.
2. To process the given slide prepared by human lymphocyte culture technique for banding.
3. To prepare the chemicals required for DNA extraction from blood samples by salting out method.
4. To carry out DNA extraction by salting out method.
5. To prepare the chemicals required for DNA extraction from buccal cells by inorganic method.
6. To carry out DNA extraction from buccal cells by inorganic method.
7. To carry out agarose gel electrophoresis for extracted DNA.
8. To study principle, working of PCR and carry out amplification for selected gene polymorphism.
9. To identify the provided photograph and interpret the type of genetic disease:
   i. Graves disease
   ii. Edward syndrome
   iii. Arthritis
   iv. Cleft lip
   v. Cleft palate
   vi. Neural tube defects
   vii. Fragile X syndrome
   viii. Hemophilia
10. To prepare karyotype for the provided metaphase plates and identify the genetic condition:
    i. Down' syndrome
    ii. Turner syndrome
    iii. Klinefelter syndrome
    iv. Patau syndrome
    v. Fragile X syndrome
11. To prepare chemicals required for conducting FISH on human chromosomes.
12. To process the given slides for FISH.
13. To interpret the FISH signals

M.Sc. Human Genetics

3rd Semester (CBCS)

Syllabus for the examination to be held in December 2023, 2024, 2025

Course No. PSHGTC-307 **Course Title: Medical Genetics and Disorders**

Credits: 04 **Maximum Marks: 100**

Time Duration: 3.0 Hrs **Minor Test I : 20**

Minor Test II : 20

Major Test : 60

Course outcomes (CO)

CO-1: The course has been designed to provide knowledge to the students of Human Genetics about the importance of Genetics in medicine, various human mitochondrial diseases, study of human genetic diseases using animal model.

CO-2: Students will be taught inheritance patterns, of different genetic diseases.

CO-3 This course will make the students to learn about the management of human genetic diseases

CO-4 Course will help the students to have knowledge about cancer, various cancer biomarkers and their role in therapeutics.

CO-5 Students will earn about genetic valuation and treatment of human infertility.

Unit-I

Medical Genetics **(12 hrs)**

1.1 Historical development of Medical Genetics.
1.2 Impact of genetics in medicine.
1.3 General concepts of disease and homeostasis.
1.4 Principles of disease diagnosis.
1.5 Epigenetics and human diseases.
1.6 Personalized Medicine: race and genetics.
1.7Nuclear transplantation and its medical implications
1.8 Synthetic biology and its applications

Unit-II

Studying Inheritance patterns **(13 hrs)**

2.1 Inheritance pattern of genetic diseases
2.1.1Autosomal dominant disorders: Huntington Disorder and Marfan syndrome.
2.1.2 Autosomal Recessive disorders: Thalasemia Sickle cell anemia, Cystic Fibrosis
2.1.3 X-linked dominant disorders: Familial rickets, Hereditary nephritis, Fragile x syndrome
2.1.4 .X- linked recessive disorders: Color blindness , Muscular dystrophies- BMD & DMD
2.2 Hemoglobin and Hemoglobinopathies
2.2.1 Structure of hemoglobin
2.2.2 Genetic control of hemoglobin synthesis
2.2.3 Developmental control of globin gene
2.2.4.Gene mutation and related abnormalities of hemoglobin
2.3. Mitochondrial diseases: Mitochondrial Myopathy, Diabetes mellitus and deafness (DAD), Leber's hereditary optic neuropathy (LHON), Myoclonic epilepsy with ragged red fibers (MERRF syndrome)

Unit-III **(12 hrs.)**

Managing Genetic Diseases

3.1 Prenatal testing
3.2 Pre-implantation of genetic diagnosis
3.3 Detection of genetic diseases
3.4 Treatment of genetic diseases
3.5 Management of genetic diseases
3.6 Consanguinity and its Consequences

M.Sc. Human Genetics

3rd Semester (CBCS)

Syllabus for the examination to be held in December 2023, 2024, 2025

Course No. PSHGTC-307 **Course Title: Medical Genetics and Disorders**

Credits: 04 **Maximum Marks: 100**

Time Duration: 3.0 Hrs **Minor Test I : 20**

Minor Test II : 20

Major Test : 60

Unit-IV **(12hrs)**

Cancers

4.1Cancer and cell cycle checkpoints
4.2 Tumor progression: Angiogenesis and metastasis
4.3DNA Repair genes and genetic instability
4.4 Cancer and Environment: Physical, chemical and biological carcinogens.
4.5 Cancer biomarkers
4.6 Genetic basis of hereditary cancers Retinoblastoma, Wilms' tumor, colorectal cancer, breast cancer.
4.7 Genomic Imprinting in cancers.
4.8 Molecular diagnostics and therapeutic interventions in cancers.

Unit-V **(13hrs)**

Genetic Basis of Infertility

5.1 Cytogenetic of male and female infertility
5.1.1 Introduction
5.1.2 Spermatogenesis
5.1.3 Oogenesis
5.2 Overview of infertility (non genetic)
5.2.1 Male
5.2.2 Female
5.3 Genetic evaluation of the
5.3.1 Infertile male
5.3.2 Infertile female
5.4 Treatment of infertilities
5.5 Assisted Reproductive Technologies

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1.5Hrs	20
Minor Test-II	21-40%	1.5Hrs	20
Major Test	41-100%	3.0 Hrs	60

i. Major test will have two sections (A&B)
ii. Section A is compulsory comprising of 10 questions of 1.5 marks each and be spread over entire syllabus
iii. Section B comprises of 6 questions (2 from each unit) from the remaining 3 units and candidate has to attemptone question from each unit (15 marks each).

Books Recommended:

1. F Vogel A.G. Motulusky. Human Genetics: Problems and Approaches. 5th Edition, BMC, 2010.
2. Helen M Kingston, ABC of Clinical genetics, , 4th Edition, BMJ, 2015.
3. Robert Nussbaum et al. Thompson & Thompson genetics in Medicine, 8th Edition, Elsevier, 2015.
4. Micheal R. Cummings Human Heredity: Principles and Issues; 11th edition, 2016,.
5. Emerys & Rimoin, Principles & Practice of Medical Genetics, 7th Edition, Elsevier, 20

M.Sc. Human Genetics

3rd Semester (CBCS)

Syllabus for the examination to be held in December 2023, 2024, 2025

Course No. PSHGTC-308 **Course Title: Medical Biotechnology with Nanotechnology**

Credits: 04 **Maximum Marks : 100**

Time Duration: 3.0Hrs **Minor Test I : 20**

Minor Test II : 20

Major Test : 60

Course outcomes (CO)

CO-1: The course will provide an opportunity to understand various diagnostics techniques like prenatal, biochemical, and Immunodiagnostic techniques.

CO-2: Course has been designed to impart knowledge of stem cells and its applications to the students.

CO-3: The course design will be focused on application of medical biotechnology in synthesis of different vaccinesand peptide based drugs.

CO-4: This course is about Introduction to Nanotechnology.

CO-5: The students will learn the principles and applications of nanotechnology.

Unit-I Diagnosis **(12 hrs)**

1.1 Prenatal diagnostics

1.1.1 Invasive techniques- Amniocentesis, Fetoscopy, Chorionic Villi Sampling

1.1.2 Non- invasive techniques- Ultrasonography, maternal fetal serum and fetal cells in the maternal blood, NIPT

1.2. Biochemical diagnostics

1.3 Immunodiagnostics

1.4 Introduction NGS to Diagnostic Course.

1.4.1 Next generation Sequencing

1.4.2 Exome sequencing

1.4.3 Whole Exome Sequencing

1.4.4 Point of Care devices (POC)

Unit-II

Stem cells and their applications **(12 hrs)**

2.1 Embryonic and adult stem cells

2.2 Characteristics of stem cell: Totipotent cells, Pluripotent cells, Multipotent cells

2.3 Culture of Stem cells

2.4 Human cord blood stem cells

2.5 Potential use of stem cells- cell based therapies

2.5.1Current treatments

2.5.2 Potential treatments

Unit-III

Applied Medical Biotechnology **(13 hrs.)**

3.1 Gene products in medicine

3.2 Anti- hemophilic factor (AHF)

3.3 Humulin

3.3.1 3 D printing / organ printing.

3.4 Erythropoietin

3.4.1 Growth hormone/ somatostatin

3.4.2 Interferon

3.4.3 DNA and RNA based vaccines

3.5 Subunit vaccines- hepatitis B vaccine

3.5.1 Attenuated vaccines

3.5.2 Peptide based drugs

M.Sc. Human Genetics

3rd Semester (CBCS)

Syllabus for the examination to be held in December 2023, 2024, 2025

Course No. PSHGTC-308 **Course Title: - Medical Biotechnology with Nanotechnology**

Credits: 04 **Maximum Marks : 100**

Time Duration: 3.0 Hrs **Minor Test I : 20**

b. Minor Test II : 20

c. Major Test : 60

Unit–IV

Introduction to Nanotechnology **(12 hrs)**

4.1 Introduction to History of nanotechnology.
4.2 Types of Nanomaterials and their Classifications.
4.3 Types of Nanocrystals-One Dimensional (1D)-Two Dimensional (2D) -Three Dimensional (3D)
4.4 Basic properties of Nanomaterials-mechanical, thermal, optical and electrical properties.
4.5 Cellular Nanostructures; Nanopores; Biomolecular motors; Bio-inspired Nanostructures

Unit- V **(13 hrs)**

Applications of nanotechnology

5.1 Nanotechnology in food industry and food packaging.
5.2 Nanotechnology in Agriculture,
5.3 Precision farming, Smart delivery system
5.4 Nanotechnology in health, drug delivery, biomedical applications.
5.5 Overview of Nanotoxicology
5.6 Nanotechnology application in energy and environment.
5.7 Gene therapy and Nanotechnology.

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1.5 Hrs	20
Minor Test-II	21-40%	1.5Hrs	20
Major Test	41-100%	3.0 Hrs	60

i. Major test will have two sections (A&B)

ii. Section A is compulsory comprising of 10 questions of 1.5 marks each and be spread over entire syllabus

iii. Section B comprises of 6 questions (2 from each unit) from the remaining 3 units and candidate has to attemptone question from each unit (15 marks each).

Books Recommended:

1. P. Nallar et al., Medical biotechnology, Oxford Handbooks, 2010.
2. Tom Strachen , Human Molecular Genetics, 4th Edition, Garland Science, 2010.
3. Surendra Nimesh, Gene therapy: Potential application of Nanotechnology, Ist Edition, Woodhead, 2013.
4. Emerys & Rimoin, Principles & Practice of Medical Genetics, 7th Edition, Elsevier, 2017.
5. Yui-Wing Francis Lam et al., Pharmacogenomics: Challenges & opportunities in TherapeuticImplementation, 2nd Edition, Academic Press, 2018.
6. Nano: The Essentials, T.Pradeep. Tata McGraw Hill, New Delhi, 2007.
7. Bharat Bhusan, “Springer Handbook of Nanotechnology”, springer, Newyork, 2007.
8. Instrumental Methods of Analysis, Willard. Merritt, Dean & Settle, CBS Publications,6th Edition, 2000.
9. Nalwa HS. 2005. Handbook of Nanostructured Biomaterials and Their Applications in Nanobiotechnology. American Scientific Publ.
10. Niemeyer CM & Mirkin CA. 2005. Nanobiotechnology. Wiley Interscience.
11. Vinod Saharan and Ajay Pal 2016. Chitosan Based Nanomaterials in Plant Growth and Protection,springerbrief

M.Sc. Human Genetics

3rd Semester (CBCS)

Syllabus for the examination to be held in December 2023, 2024, 2025

Course No. PSHGTC- 309 **Course Title: - Evolution and Phylogenetics**

Credits: 02 **Maximum Marks : 50**
Time Duration: 2.5Hrs **Minor Test I : 05**
Minor Test II : 05
Major Test : 40

Course outcomes:

CO-1: The course is designed with the objective to provide knowledge to the students about the concept and theories of evolution

CO-2: The students will have an understanding of genetic constitution of a population.

CO-3: The students will learn the concepts of population genetics; phenotype and genotype analysis.

Unit-I

Evolution **(10 hrs)**

1.1 Concept and Theories of Evolution
1.2 Natural selection
1.3 Modes of Speciation
- 1.3.1 Allopetric , parapetric and sympatric speciation
- 1.3.2 Evolutionary processes causing speciation
- 1.3.3 Natural Selection
- 1.3.4 Sexual Selection

1.4 Random genetic drift, muller incompatibility, gene pool, genetic migration, geographical isolation. Haplotype mapping,
1.5 Molecular Phylogeny
- 1.5.1 Phylogenic analysis
- 1.5.2 Construction of Phylogenetic trees.
- 1.5.3 Introduction to Comparative Genomics

Unit-II

Genetic constitution of a population **(10hrs)**

2.1 Genetic constitution of a population
2.2 Gene frequencies and genotypes.
- 2.2.1 Hardy Weinberg Law
- 2.2.2 Testing of HWE in the population
- 2.2.3 Impact of recurrent and non recurrent mutations in HWE population

2.3 Changes in gene frequency and continuous variation -Environmental variance.
2.4 In-breeding depression & mating systems, migrations, adaptive landscape, spatial variation & genetic fitness.

Unit-III

Population Genetics **(10 hrs.)**

3.1 Phenotype and Genotype analysis
3.2 Human Migration and diseases: Founder effect, bottle neck effect, genetic effect.
3.3 Type of genetic association studies
- 3.3.1 Family based studies
- 3.3.2 Population based studies
- 3.3.3 Genome wide Association Studies (GWAS)
- 3.3.4 Genomic selection
- 3.3.5 Cross Sectional studies

3.5 Genetic Polymorphism and Inherited variations.
3.6. Bayesian statistics, adapted landscape, special variations and genetic fitness.

M.Sc. Human Genetics

3rd Semester (CBCS)

Syllabus for the examination to be held in December 2023, 2024, 2025

Course No. PSHGTC- 309 **Course Title: - Evolution and Phylogenetics**

Credits: 02 **Maximum Marks : 50**

Time Duration: 2.5Hrs **Minor Test I : 05**

Minor Test II : 05

Major Test : 40

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	5
Minor Test-II	21-40%	1 Hr	5
Major Test	41-100%	2.5Hrs	40

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

Books Recommended:

1. Dummies, Jean-Michel Claverie, Cedric Notredame, Bioinformatics, John Wiley & Sons, 2003.
2. John H Relethford, Human Population genetics, Wiley Blackwell, 2011.
3. Robert Palo Min, Behavioral Genetics, 6th Edition, Worth Publishers, 2012.
4. Arthur M. Lesk, Introduction to Bioinformatics, 4th Edition, Oxford, 2013.
5. Robert Nussbaum et al. Thompson & Thompson genetics in Medicine, 8th Edition, Elsevier, 2015.
6. Lisa M. Sullivan, Essentials of Biostatistics in Public Health, 3rd Edition, Jones & Bartlett Learning, 2017.
7. Jennifer Doudna, Crack in Creation: the new power to control evolution, Vintage Digital, 2017.

M.Sc. Human Genetics

3rd Semester (CBCS)

Syllabus for the examination to be held in December 2023, 2024, 2025

Course No. PSHGTC- 310 **Course Title: - Bioinformatics and Biostatistics**

Credits: 02 **Maximum Marks : 50**

Time Duration: 2.5Hrs **Minor Test I : 05**

Minor Test II : 05

Major Test : 40

Course outcomes:

CO-1: The course is designed with the objective to provide knowledge to the students about the scope of bioinformatics

CO-2: The students will lean the use of various bioinformatics tools.

CO-3: This course will make the students to learn the concept and methods of Biostatistics.

Unit-I **(10 hrs)**

Bioinformatics

1.1 Scope of Bioinformatics.

1.2 Biological Databases; Primary, Secondary & Composite databases, UCSC Genome brower.

1.3 Nucleotide Sequence Databases; GenBank, ENSEMBL, DDBJ

1.3 Protein Sequence Databases; SWISS-PROT, protein sequence database, Translated EMBL (TrEMBL), UniProt, PROSITE, Pfam, KEGG pathway

1.4 Structural Databases; Protein Data Bank (PDB), Molecular Modelling Database (MMDB), Nucleic Acid Database (NDB).

1.5 Expression data set

1.5.1 Single cell gene expression

1.5.2 GTEX, ENCODE.

1.6 Clinical Database: Gene cards, OMIM

Unit-II **(10 hrs)**

Bioinformatics

2.1 Retrieval Systems: SRS (Sequence Retrieval System) for flat file format libraries, ENTREZ, Global Query for NCBI Search, DBGET/ LinkDB: Database of link information.

2.2 Sequence Similarity Search: BLAST, FASTA, CLUSTALW.

2.3 Sequence submission tools: Bankitt, Sequin, Webin, SAKURA.

2.4 Sequence assembly, Genome analysis, Phylogenetic analysis.

2.5 Comparative genomics: cluster of orthologous groups (COGs), Homologene at NCBI

Unit- III **(10 hrs)**

Biostatistics

3.1 Measures of central tendency and measures of dispersion, probability and its types: permutation, combination, probability computations.

3.2 Theoretical distributions: Binomial, Poisson and Normal, hypothesis testing; two types of errors.

3.3 Tests of significance; Z-test, t-test, chi-square test, one way and two way analysis of variance.

3.4 Simple correlation and regression.

3.5 Hypothesis and Testing : Statistic and Parameters, Population and sample size, Null and alternative hypothesis, Odds Ratio.

M.Sc. Human Genetics

3rd Semester (CBCS)

Syllabus for the examination to be held in December 2023, 2024, 2025

Course No. PSHGTC- 310 **Course Title: - Bioinformatics and Biostatics**

Credits: 02 **Maximum Marks : 50**

Time Duration: 2.5Hrs **Minor Test I : 05**

Minor Test II : 05

Major Test : 40

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	5
Minor Test-II	21-40%	1 Hr	5
Major Test	41-100%	2.5Hrs	40

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

Books Recommended:

1) Lesk, A. M. (2002). Introduction to Bioinformatics. Oxford: Oxford University Press.
2) Mount, D. W. (2001). Bioinformatics: Sequence and Genome Analysis.Cold Spring Harbor, NY: Cold Spring Harbor Laboratory Press.
3) Baxevanis, A. D., & Ouellette, B. F. (2001). Bioinformatics: a Practical Guide to the analysis of Genes and Proteins. New York: Wiley-Interscience.
4). Pevsner, J. (2015).Bioinformatics and Functional Genomics. Hoboken, NJ.: Wiley- Blackwell.
5) Bourne, P. E., & Gu, J. (2009). Structural Bioinformatics. Hoboken, NJ: Wiley-Liss.
6). Lesk, A. M. (2004). Introduction to Protein Science: Architecture, Function, and Genomics. Oxford: Oxford University Press.
7). Baxevanis, A.D. and Francis Onellete, B.F. (2001). Bioinformatics. Wiley Interscience. John Wiley and Sons Inc. New York.
8). Attwood, T.K. and Parry-Smith, D.J. (1999). Introduction to Bioinformatics. Pearson Education Ltd., Singapore.

M.Sc. Human Genetics

3rd Semester (CBCS)

Syllabus for the examination to be held in December 2023, 2024, 2025

Course No. PSHGTO-311 **Course Title: - IPR AND BIOETHICS**

Credits: 02
Time Duration: 2.5Hrs

Maximum Marks : 50
Minor Test I : 05
Minor Test II : 05
Major Test : 40

Course Outcomes:

CO-1: The course is designed with the objective to provide knowledge to the students about role of intellectual property rights in genetics.

CO-2: Students will be briefed about the principles of ethics.

CO-3: This course is designed with the objective to provide knowledge about entrepreneurship in genetics.

Unit—I (10hrs)

Intellectual Property Rights

1.1 Role of IPRs in genetics, types of IPRs, patent protection in the constitutions, Purpose of a Patent.
1.2 Material transfer Agreements, Promoting Technological Advancement.
1.3 Patentable Inventions, Patent Requirements.
1.4 Patent Application, Patenting organisms, Patent Licensing
1.5 Research and IP, Major treaties that govern IP: Introduction to Indian and US patent offices.
1.6 TRIP'S and various provisions in the TRIPS Agreement, Benefits of securing IPRs; Indian legislations for the protection of various types of IPs; National Biodiversity protection initiatives.

Unit-II (10hrs)

BIOETHICS

2.1 Traditional knowledge and bioethics.
2.2 Bioactivities.
2.3 Ethical Issues, Statement of Bioethical Principles
2.4 Medical Privacy, Confidentiality and Genetic Discrimination, Informed Consent.
2.5 Stem Cells, Cloning.
2.6 Eugenics
  2.6.1 Euphenics and Euthenics, Euthanasia.
  2.6.2. Human Cloning and Eugenics
2. 7 Basic ethic principles: autonomy, beneficence, Non-maleficence, justices, integrity.

Unit-III (10hrs)

Entrepreneurship in genetics based industries

Introduction to social and business entrepreneurship; basic characteristics, developing entrepreneurship through training and motivation.

3.2 Concept of enterprise, importance of spotting the opportunity leveraging resources and creating value.
3.3 Customer needs and market segmentation; categories of value: enhancements, extensions and specializations.
3.4 Entrepreneurial opportunities in genetics: Structure of the industry: companies working in different areas of specialization and different stages (R and D, manufacture, sales and marketing) industry: policy making relevant to genetics.
3.5 Drug designing

M.Sc. Human Genetics

3rd Semester (CBCS)

Syllabus for the examination to be held in December 2023, 2024, 2025

Course No. PSHGTO-311 **Course Title: - IPR AND BIOETHICS**

Credits: 02
Time Duration: 2.5Hrs

Maximum Marks : 50
Minor Test I : 05
Minor Test II : 05
Major Test : 40

Note for paper setting:

Examination theory weightage	Syllabus to be coveredin examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	5
Minor Test-II	21%1-40%	1 Hr	5
Major Test	41%-100%	2.5 Hrs	40

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, 6 th 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.
10. M.B. Rao; (2008) Biotechnology, IPRs and biodiversity, Pearson Publications.

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3rd Semester (CBCS)

Syllabus for the examination to be held in December 2023, 2024, 2025

LAB COURSE NO. PSHGPC-305

List of practicals based on Theory Course no. 307, 308.

1. To study various chromosome changes in cancer with respect to ploidy changes in cervical cancer.
2. To study the presence of Micronuclei in different types of cancers as a biomarker for detecting the severityof the disease.
3. To find out the Philadelphia chromosome from the provided microphotograph.
4. To determine the blood sugar level (glucose) with the help of glucometer.
5. To prepare karyotype from the provided microphotograph of metaphase showing human chromosomeswith GTG-banding.
6. To estimate Hb from your own blood by using Hematocytometer.
7. To perform screening test for beta thalassemia (NESTROFF test).
8. To perform color blindness test by using Ishihara charts.
9. To prepare the chemicals required for DNA extraction from blood samples by organic method.
10. To carry out DNA extraction from blood samples by organic method.
11. To prepare the chemicals required for DNA extraction from blood samples by salting out method.
12. To carry out DNA extraction from provided blood samples by inorganic method.
13. To carry out DNA extraction from provided blood samples by commercial kit method.
14. To perform qualitative and quantitative analysis of extracted DNA.
15. To carry out the PCR amplification a selected gene SNP.
16. To perform restriction digestion of a selected gene SNP and study its RFLP pattern.
17. To prepare the karyotype of the Infertile couples.

LAB COURSE NO. PSHGPC-306

Credits: 04 **Max.Marks:100**

List of practicals based on Theory Course no. 309, 310 and 311.

1. To study the different symbols used in pedigree analysis and their significance.
2. To draw and interpret the pedigree of your own family.
3. To study different patterns of inheritance in humans.
4. To study the different dominant and recessive traits in humans:
    a. Tongue rolling mechanisms
    b. Eye colour
    c. Widow's peak
    d. PTC tasting
5. Study of Hardy Weinberg equilibrium in a given population group.
6. To construct and study phylogenetic tress.
7. To study fingerball and palmar dermatoglyphics and calculate indices.
8. Bioinformatics practical sessions based on : Nucleic acid databases, Genome databases, Protein databases
9. Biostatistics analysis exercises based on
    a. Calculation of central tendencies
    b. T-test
    c. Chi square test
    d. Odd ratio
    e. ANOVA
    f. Z-test g. F-test h. Mean i. Standard deviation

M.Sc. Human Genetics

4th Semester (CBCS)

Syllabus for the examination to be held in May 2024, 2025, 2026

Course No. PSHGTC-406
Credits: 04
Time Duration: 3.0Hrs

Course Title: - Genetic Counseling
Maximum Marks : 100
Minor Test I : 20
Minor Test II : 20
Major Test : 60

Course Outcomes

CO-1: The course has been designed with the objective to make students learn about the importance of Genetic Counseling in the welfare of family as well as society.

CO-2: It will also help in providing knowledge to students that how genetic counseling services can be used to detect a genetic condition.

CO-3: Course will provide an insight into different testing issues viz. discrimination, privacy and confidentiality.

CO-4: The course will further help in tacking the issues associated with human cloning, organ transplantation &Surrogacy. Students will also gather information regarding ethics involved in medicine in Indian system.

CO-5: The course will provide a comprehension picture about relation of Pre-conception and Pre-natal Diagnosis Act in Human Genetics.

Unit-I

Genetic Counseling **(12 hrs)**

1.1Introduction to Genetic counseling
- 1.1.1. Scope of Genetic counseling
- 1.1.2. Genetic counseling in Mendelian disorders and non- Mendelian disorders
- 1.1.3Types of Genetic counseling: Individual, Family and Group

1.2 Genetic counselors: Definition, role, qualities and responsibilities
1.3 Process of Genetic Counseling
1.4 Psychosocial and behavioral aspects of Genetic Counseling
1.5 Pedigree Charting: Essential for Genetic Counseling.
1.6 Ethos of Genetic services and genetic counseling, Indications of Genetic counseling

Unit-II

Genetic counseling and genetic disorders **(13 hrs)**

2.1 Neuromuscular diseases
2.2 Infertility
2.3 Hereditary Cancers
2.4 Trinucleotide repeat expansions
2.1 Disorders of bone and connective tissues
2.2 Oral and craniofacial disorders
2.3 Deafness and renal diseases

Unit-III **(12 hrs)**

Issues in Genetic testing-I

3.1. Genetic testing issues
- 3.1.1 Privacy and Confidentiality
- 3.1.2. Genetic Discrimination
- 3.2.3. Issues in X linked Diseases.

3.2. Maintenance of genetic register
3.3 Genetic counseling clinics and its working
3.4 How to get ethical approval from management.
3.5 Status of Genetic Counseling in India and Abroad.

M.Sc. Human Genetics

4th Semester (CBCS)

Syllabus for the examination to be held in May 2024, 2025, 2026

Course No. PSHGTC-406 **Course Title: - Genetic Counseling**

Credits: 04 **Maximum Marks : 100**

Time Duration: 3.0Hrs **Minor Test I : 20**

Minor Test II : 20

Major Test : 60

Unit-IV **(12 hrs)**

Issues in Genetic testing-I

4.1 Informed Consent and Right of Choice

4.2 Human Cloning and Eugenics

4.3 Surrogate mothers

4.4 Organ banking and transplantation

4.5 Medical Ethics in India

4.6 Dilemmas faced by counselors

Unit-V

Legal Implications in Genetic Testing **(13 hrs)**

5.1. Pre-natal diagnostic techniques (Regulation and Prevention of Misuse) Act, 1994

5.1.1 Pre conception Pre-natal diagnostic techniques (Prohibition of sex selection) Act

5.2 Regulation of prenatal diagnostic techniques

5.3 Registration & regulation of genetic counseling centers, genetic laboratories & genetic clinics

5.4 Appropriate authority & advisory committee

5.5 Offences and Penalties

5.6 Medical termination of pregnancy Act

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1.5Hrs	20
Minor Test-II	21%-40%	1.5Hrs	20
Major Test	41%-100%	3.0 Hrs	60

i. Major test will have two sections (A&B)
ii. Section A is compulsory comprising of 10 questions of 1.5 marks each and be spread over entire syllabus
iii. Section B comprises of 6 questions (2 from each unit) from the remaining 3 units and candidate has to attempt one question from each unit (15 marks each).

Books Recommended:

1. Rimion et al., Principles and Practice of Medical Genetics, Vol-I-III, Churchill, 2002
2. Christine Evans, Genetic Couselling: a psychological approach, Cambridge University Press, 2006
3. Young, Introduction to Risk Calculation in Genetic Counselling, 3rd Edition, Oxford, 2007.
4. Susan Schmerler, Lessons learned: Risk Management issues in Genetic Counseling, Springer, 2008.
5. M. Fox., A guide to Genetic Counseling, 2nd Edition, Elsevier, 2010.
6. Vandana Mudda, PC&PNDT Act, Blackwells, 2012.
7. Janice L. Berliner, Ethical Dilemas in Genetics & genetic counseling, Oxford University Press, 2014.
8. McKinsey L. Goodenberg et al., Practical genetic counseling for the laboratory, Oxford, 2017.

M.Sc. Human Genetics

4th Semester (CBCS)

Syllabus for the examination to be held in May 2024, 2025, 2026

Course No. PSHGTC-407 | **Course Title: - Clinical Genetics**

Credits: 04 | **Maximum Marks : 100**

Time Duration: 3.0Hrs | **Minor Test I : 20**

Minor Test II : 20

Major Test : 60

Course Outcomes:

CO-1: The course has been designed with the objectives to make the students to learn about the role of clinical genetics.

CO-2: The course will provide knowledge on reprogenetics.

CO-3: Students will able to get information on treatment of genetic diseases.

CO-4: The course will give an elaborative insight on etiology and genetics of multifactorial diseases.

CO-5: The course will provide an understanding on rare genetic diseases.

Unit-I **(12hrs)**

Principles and practice of clinical genetics

1.1 Molecular and Biochemical basis of genetic diseases

1.2 Clinical utilization of presymptomatic and predisposition testing

1.3 Presymptomatic testing for genetic diseases and malignancy.

1.4 Late onset genetic disorders- Alzheimer, Parkinson's disease, Schizophrenia

1.5 Nature and Nurture: Distangling the effects of genes and environment

Unit-II **(13hrs)**

Abnormality and Disorders.

2.1 Implications of Chromosome abnormalities on human health

2.2 Genetics of Pregnancy loss

2.3 Ring chromosome and related genetic disorders

2.4 Clinical significance of chromosome, DNA and foetal imaging in the context of fetal abnormality

2.5 Reprogenetics- Germinal Choice Technology

Unit-III **(12hrs)**

Diagnosis and Management.

3.1. Treatment of genetic diseases

3.1.1. Conventional approaches to the treatment of genetic diseases

3.2. Genetic screening and genetic testing, Newborn screening, population carrier screening

3.3.Clinical basis of genetic diagnostics

3.4.Therapeutic aspects and emerging therapies of genetic diseases

3.5. Fetal treatment

3.6.Introduction to personalized medicine

Unit-IV **(13 hrs)**

Multifactorial Disorders.

4.1 Introduction to Multifactorial diseases

4.2 Examples of multifactorial diseases

4.2.1 Cardiovascular diseases

4.2.2 Hyperthyroidism

4.2.3 Obesity

4.3 Estimation of genetic component of multifactorial traits: empiric risk score.

4.4 Heritability; coefficient of relationship

4.5 Application of Bayes Theorem

4.6 MCG Care guidelines.

M.Sc. Human Genetics

4th Semester (CBCS)

Syllabus for the examination to be held in May 2024, 2025, 2026

Course No. PSHGTC-407 **Course Title: - Clinical Genetics**
Credits: 04 **Maximum Marks : 100**
Time Duration: 3.0Hrs **Minor Test I : 20**
Minor Test II : 20
Major Test : 60

Unit-V **(13 hrs)**

Rare Genetic Diseases

5.1. Introduction to rare genetic diseases
5.2. Lysosomal storage diseases
5.3. Neurofibromatosis
5.4. Progeria
5.5. Werewolf syndrome
5.6. Skeletal dysplasia

Note for paper setting:

Examination theory weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1.5 Hrs	20
Minor Test-II	21-40%	1.5Hrs	20
Major Test	41-100%	3.0 Hrs	60

i. Major test will have two sections (A&B)
ii. Section A is compulsory comprising of 10 questions of 1.5 marks each and be spread over entire syllabus
iii. Section B comprises of 6 questions (2 from each unit) from the remaining 3 units and candidate has to attempt one question from each unit (15 marks each).

Books Recommended:

1. D. Peter Snustad and Michael J.Simmons. Principles of Genetics. 6th edition. John Wiley & Sons, Inc., 2011.
2. ABC of Clinical genetics, Helen M Kingston, 4th Edition, BMJ, 2015.
3. Human Heredity: Principles and Issues by Micheal R. Cummings; 11th edition, Cengage Learning, 2016,.
4. Drs. Peter Turnpenny and Sian Ellard., Emery's Elements of Medical Genetics, 15th edition , Elsevier, 2017.
5. Emerys & Rimoin, Principles & Practice of Medical Genetics, 7th Edition, Elsevier, 2017.

M.Sc. Human Genetics

4th Semester (CBCS)

Syllabus for the examination to be held in May 2024, 2025, 2026

Course Title: - Human Genetic Disorders & Society **Course No. PSHGTO-408**
Credits: 04 **Maximum Marks : 100**
Time Duration: 3.0Hrs **Minor Test I : 20**
Minor Test II : 20
Major Test : 60

Course Outcomes

CO-1: The course has been designed to expose the students to the present state about the field of molecular genetics & biotechnology.
CO-2: Students will able to have knowledge on single gene disorders.
CO-3: The course will help the students to learn about the different techniques used in detecting genetic abnormalities.
CO-4: Students will able to get understanding about genetics of multifactorial diseases.
CO-5: The course will also help the students to learn about the importance of Genetic Counseling in the welfare of family as well as society.

Unit-I **(12hrs)**

1.1 Chromosomes and cell division
1.2 Structure and functions of chromosomes
1.3 Human chromosomes
1.4 Cell division:
1.4.1 Mitosis
1.4.2 Meiosis
1.5 Analysis of human chromosomes
1.6 Conventional techniques
1.7 Advanced techniques

Unit-II **(12hrs)**

2.1 Central Dogma of Life
2.2 DNA as a genetic material
2.3 DNA Replication
2.4 Transcription
2.5Translation

Unit-III **(13hrs)**

3.1 Genetic Disorders
3.2 Chromosomal Disorders: An introduction
3.3 Trisomies: Down syndrome, Edward syndrome, Patau Syndrome, Turner Syndrome, Klinefelter syndrome
3.4 Single Gene Disorder: Sickle Cell Anemia, Huntington Disease.
3.5 Complex Disorders
3.5.1 CVD
3.5.2 DM
3.5.3 Schizophrenia
3.5.4 Breast Cancer
3.5.5 Leukemia

Unit-IV **(13hrs)**

4.1 Molecular Genetic techniques
4.2 PCR: principle, working and its applications.
4.2.1 Electrophoresis (Overview)
4.2.2 FISH
4.2.3 DNA fingerprinting
4.3 DNA Sequencing
4.3.1 Chain termination method
4.3.2 Chemical degradation method

M.Sc. Human Genetics

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Major Test : 60

Unit-V **(13hrs)**

Genetic Counseling

5.1 Genetic counseling: an Overview.

5.2 Goals of Genetic Counseling.

5.2.1 Types of Genetic Counseling.

5.2.2 Traits and Credentials of Genetic Counselor.

5.3 Breaking a bad news

5.4 Pedigree Charting: Essential for Genetic Counseling.

5.5 Ethos of Genetic services and genetic counseling

5.6. Indications of Genetic Counseling

5.7 Psychosocial and Behavior aspect of Genetic Counseling

Note for paper setting:

Examination theory Weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1.5Hrs	20
Minor Test-II	21%-40%	1.5Hrs	20
Major Test	41-100%	3.0 Hrs.	60

i. Major test will have two sections (A&B).
ii. Section A is compulsory comprising of 10 questions of 1.5 marks each and be spread over entire syllabus
iii. Section B comprises of 6 questions (2 from each unit) from the remaining 3 units and candidate has to attempt one question from each unit (15 marks each).

Books Recommended:

1. D. Peter Snustad and Michael J.Simmons. Principles of Genetics. 6th edition. John Wiley & Sons, Inc., 2011.
2. Vandana Mudda, PC&PNDT Act, Blackwells, 2012.
3. Janice L. Berliner, Ethical Dilemas in Genetics & genetic counseling, Oxford University Press, 2014.
4. Helen M Kingston, ABC of Clinical genetics, , 4th Edition, BMJ, 2015.
5. Emerys & Rimoin, Principles & Practice of Medical Genetics, 7th Edition, Elsevier, 2017.
6. Drs. Peter Turnpenny and Sian Ellard., Emery's Elements of Medical Genetics, 15th edition , Elsevier, 2017.

M.Sc. Human Genetics

4th Semester (CBCS)

Syllabus for the examination to be held in May 2024, 2025, 2026

Credits: 04 **Max.Marks:100**

LAB COURSE NO. PSHGPC-405

List of practical's based on Theory Course no. 406 & 407

1. How to take clinical history of a suspected/patient/client.
2. To study the communication process of genetic counseling for genetic testing.
3. Designing proforma for different genetic diseases.
4. Prenatal screening questionnaire design.
5. Pre-conceptional screening and counseling
6. To process the given slide prepared by human lymphocyte culture technique for banding.
7. To prepare chemicals required for GTG banding.
8. To prepare the chemicals required for DNA extraction from blood samples by organic method.
9. To carry out DNA extraction from blood samples by organic method.
10. To carry out agarose gel electrophoresis for extracted DNA.
11. To re-precipitate DNA from provided stored DNA samples.
12. To standardize PCR conditions and carry out PCR amplification of the given gene.
13. To perform gel elution of separated PCR products from agarose gel.
14. Case studies of different genetic disorders/syndromes.
15. Management of different genetic disease