INSTITUTE OF HUMAN GENETICS

M. Sc. Human Genetics

Program Outcome (PO)

PO-1 **Knowledge:** The degree in M. Sc .Human s Genetics offers knowledge and about the 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 emphasises in order to make 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. The students can conduct surveys regarding the identification of various genetic disorders.

PO-4 **Analytical based studies with result outcome:** After understanding principals 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 the relevant 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 so analyzed can be implemented towards the improvement of diagnosis and treatment of diseases. The data so calculated can be added to the state 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 in the 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 counselling and diagnosis.

M.Sc. Human Genetics

Ist Semester (CBCS)

Syllabus for the examination to be held in December, 2019, 2020 and 2021

Course No. PSHGTC-101 **Course Title: - Cell Biology**

Credits: 04 **MAXIMUM MARKS : 100**

Time Duration: 2 Hrs & 30 mins **a. Minor Test I : 20**

b. Minor Test II : 20

c. Major Test :60

Course Outcomes (CO)

CO-1: -The course has been designed to expose the students of Human Genetics to understand the structure and function of cell membrane and related transport mechanisms,

CO-2: Students will be able to understand structure, function of different cell organelles, bio molecules,

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

Cell: The basic unit of life **(13 hrs.)**

1.1. Cell: Structure and Organisation

1.2. Plasma Membrane

1.2.1. Structure of Plasma Membrane with special emphasis on various models

1.2.1. Functions of Plasma Membrane

1.2.2.1. Transport across membrane

1.2.2.2. 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

1.3.3. Intermediate filaments

Unit-I

Understanding Cell Organelles

(12hrs.)

2.1. Mitochondria

2.1.1. Ultrastructure

2.1.2. Role of mitochondria in 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

2.3.3. Lysosomes

2.3.4. Peroxisomes

Unit-III

Cell division **(12hrs.)**

3.1. Mitosis:

3.1.1 Phases of Mitosis

3.1.2. Significance and Consequences of Mitosis

3.2. Meiosis

3.2.1 Premeiotic and Meiotic stages

3.2.2 Chromosome synapsis and Synaptonemal complex

3.2.3 Mechanism of crossing over, genetic recombination & Meiotic defects

3.2.4 Genetic Consequences of Meiosis

3.2. Meiosis

3.2.1 Premeiotic and Meiotic stages

3.2.2 Chromosome synapsis and Synaptonemal complex

3.2.3 Mechanism of crossing over, genetic recombination & Meiotic defects

3.2.4 Genetic Consequences of Meiosis

Unit-IV

Cellular interactions and Cell Cycle

4.1 Cell-Cell Interaction **(13 hrs.)**

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 Centrosome Cycle
4.2.2 Cell cycle check points
4.2.3 Role of Rb and p53 protein in cell cycle regulation

Unit-V

Cell Signaling **(12 hrs.)**

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 Adenylyl cyclase, Phophotidylinositol effectors)
5.4. Desensitization (termination of GPCR signaling)
5.5. Enzyme linked receptor signaling (RTK signaling; Ras-MAPK signaling; JAK-STAT 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 Hr	20
Minor Test-II	21-40%	1 Hr	20
Major Test	41-100%	2 Hrs.& 30 mins	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, 2019, 2020 and 2021

Course No. PSHGTC-102 | **Title: - Human Physiology**
Credits: 04 | **MAXIMUM MARKS : 100**
Time Duration: 2 Hrs & 30 mins | **a. Minor Test I : 20**
b. Minor Test II : 20
c. Major Test : 60

Course Outcomes: - Course is designed to apprise the student about the details of physiological aspects of different systems of human body. This will help the students to understand the changes caused in different systems and organs due to genetic changes.

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 its different types
- 1.2.2 Physiology of muscle contraction
- 1.2.3 Neuromuscular Junction

Unit-II

Human Systems and their Physiology –I **(12 hrs.)**

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

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

Human Systems and their Physiology –III **(13 hrs.)**

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
- 4.2.2 Thyroid, Parathyroid Gland
- 4.2.3 Adrenal Gland
- 4.2.4 Islets of Langerhans
- 4.2.5 Gonads

Unit-V

Human Embryology **(12 hrs.)**

5.1 Male & female Reproductive Systems
5.2 Mechanism of Human Fertilization
5.3 Implantation
5.4 Development of human embryo upto three germinal layers
5.5 Development of embryonic disc, notochord formation & neurulation
5.6 Chronic formation & dev. of placenta

Note for paper setting:

Examination theory weightage	Syllabus to be covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	20
Minor Test-II	21-40%	1 Hr	20
Major Test	41-100%	2 Hrs.& 30 mins	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, 2019, 2020 and 2021.

Course No. PSHGTC-103 | **Course Title: - Human Molecular Genetics-I**
Credits: 04 | **MAXIMUM MARKS : 100**
Time Duration: 2 Hrs & 30 mins | **a. Minor Test I : 20**
b. Minor Test II : 20
c. 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 and functions of Nucleic Acid.
CO-2: Understanding the concepts of Translation and gene regulation.
CO-3: Knowledge about significance and impact of mutations.
CO-4: Introduction to Basic concepts of Human Genome and its evolution.
CO-5: Understanding about the various hybridization assays.

Unit-I
Human Genome **(13 hrs.)**

1.1 Basic concepts of Human Genome
1.2 Human Gene Families
1.3 Homology, Paralogs & Orthologs
1.4 Repetitive DNA and its types
1.5 Transposable elements
1.6 Genome evolution
- 1.6.1 Nuclear genome evolution
- 1.6.2. Mitochondrial genome evolution
- 1.6.3. Sex chromosome evolution

Unit-II
Nucleic Acid : Structure & functions **(13 hrs.)**

2.1 DNA – Structure & types
2.2 DNA Replication in Prokaryotes & Eukaryotes.
2.3 RNA – Structure & types
2.4 Mechanism of Transcription and transcription factors
2.5 Post-transcriptional modification:
- 2.5.1 5'Capping
- 2.5.2 Polyadenylation
- 2.5.3 Splicing
- 2.5.4 RNA editing

Unit-III **(12 hrs.)**
Translation and Gene Regulation

3.1 Mechanism of Translation (Initiation, Elongation & Termination)
3.2 Positive and Negative Regulation (Lac Operon and Tryptophan model)
3.3 Gene Regulation in Eukaryotes
- 3.3.1 Regulation at Transcriptional level
- 3.3.2 Regulation at post-transcriptional level (RNA interference).

3.4 m-RNA degradation

Unit-IV
DNA Mutations **(12 hrs.)**

4.1. Mutations
- 4.1.1 Types of mutations
- 4.1.2 Physical & Chemical Mutagens

4.2 Ames test
4.3 Complementation test

M.Sc. Human Genetics, Ist Semester (CBCS)
C. No. PSHGTC-103, Human Molecular Genetics-I (Dec., 2019-21*)*

DNA Repair

4.4 DNA repair mechanism
- 4.4.1 Direct Repair
- 4.4.2. Excision Repair (Base Excision Repair, Nucleotide Excision Repair, Mismatch Repair, Non homologous end joining , SOS repair)

Unit-V

Molecular Hybridization **(12 hrs.)**

5.1 DNA hybridization assays
5.2 Nucleic acid probe
5.3 Principles of molecular hybridization
5.4 Methods and applications of molecular hybridization.
5.5 Synthesis and labeling of probes

Note for paper setting:

Examination theory weightage	Syllabus to be covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	20
Minor Test-II	21-40%	1 Hr	20
Major Test	41-100%	2 Hrs.& 30 mins	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 e tal, 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, 2019, 2020 and 2021

Course No. PSHGTC-104 | **Course Title: - Principles of Human Genetics**
Credits: 02 | **MAXIMUM MARKS : 50**
Time Duration: 2 Hrs | **a. Minor Test I : 10**
b. Minor Test II : 10
c. Major Test : 30

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 History of Cytogenetics
1.2 Mendel's Laws of Heredity: Law of segregation & Law of Independent Assortment
1.3 Test Cross & Back Cross
1.4 Extentions of Mendelism:
- 1.4.1 Incomplete Dominance
- 1.4.2 Co-dominance
- 1.4.3 Multiple allelism
- 1.4.4 Epistasis
- 1.4.5 Pleiotropy

1.5 Chromosomal Theory of Heredity / Inheritance and Non disjunction as a proof to Chromosomal Theory

Unit-II
Introduction to Chromosomes **(10 hrs.)**
2.1 Structure and Organization of Human Chromosomes:
- 2.1.1 Nomenclature of Chromosomes
- 2.1.2 Landmarks of Chromosomes

2.2. Structural Changes in chromosomes (Deletions, Duplications, Translocations, Inversions)
2.3 Numerical changes in chromosomes (Aneuploidy, Polyploidy)
2.4 Numerical Abnormalities of Autosomes & Sex Chromosomes: Down Syndrome, Edward Syndrome & Patau Syndrome ,Turner syndrome & Klinefelter syndrome

Unit-III
Chromosomal & Non-chromosomal basis of sex determination **(10 hrs.)**
3.1. Sex determination & differentiation in Humans
3.2 Dosage compensation- Lyon Hypothesis
3.3 Inactivation of Sex Chromosomes
3.4. Genetic Balance theory of sex determination in Drosophila
3.5. Non chromosomal basis of sex determination
3.6. Androgen Insensitivity syndrome
Note for paper setting:

Examination theory weightage	Syllabus to be covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	10
Minor Test-II	21-40%	1 Hr	10
Major Test	41-100%	2 Hrs	30

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, 2017.

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M.Sc. Human Genetics
Ist Semester (CBCS)
Syllabus for the examination to be held in December, 2019, 2020 and 2021

Course No. PSHGTC-105 **Course Title: - Genetic diagnostics & Human Health Care**
Credits: 02 **MAXIMUM MARKS : 50**
Time Duration: 2 Hrs **a. Minor Test I : 10**
b. Minor Test II : 10
c. Major Test : 30

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 different types of blotting techniques.
CO-3: Understanding various types of Chromosomal and Molecular analysis techniques.

Unit-I **(10 hrs.)**
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.Chromatography techniques: Paper chromatography, Liquid chromatography, gas chromatography, TLC.

Unit-II **(10 hrs.)**
2.1 Blotting Techniques
2.1.1. Southern Blotting
2.1.2. Northern Blotting
2.1.3. Dot Blot Assay
2.2 Microscopy
2.2.1. Light Microscopy
2.2.2 Phase-contrast Microscopy
2.2.3 Fluorescence Microscopy
2.2.4 Electron Microscopy, SEM, TEM.

Unit-III **(10 hrs.)**
3.1. Lymphocyte Culturing
3.2. Chromosome Banding Techniques
3.2.1. G-banding
3.2.2. Q-banding
3.2.3. R-banding
3.2.4. C-banding
3.2.5. NOR-banding
3.2.6. High Resolution Banding
3.3. Molecular techniques
3.3.1. Polymerase chain reaction
3.3.2. RFLP
3.3.3. DNA Sequencing
Note for paper setting:

Examination theory weightage	Syllabus to be covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	10
Minor Test-II	21-40%	1 Hr	10
Major Test	41-100%	2 Hrs	30

i. 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. 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.

M.Sc. Human Genetics
Ist Semester (CBCS)

Syllabus for the examination to be held in December, 2019, 2020 and 2021.

Note: Alterations will be done in the following listed practicals depending on the research work going on in the Major Research Lab of IHG with authorization of the Coordinator.

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. 103, 104 & 105

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.

M.Sc. Human Genetics
Ist Semester (CBCS)

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

Syllabus for the examination to be held in December, 2019, 2020 and 2021

Note: Alterations will be done in the following listed practicals depending on the research work going on in the Major Research Lab of IHG with authorization of the Coordinator.

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

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

List of practicals based on Theory Course no. 103, 104 & 105

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
    f. Down syndrome
    g. Edward syndrome
    h. Patau syndrome
    i. Turner syndrome
    j. Klinefelter syndrome
9. To study the scheme of the Karyotype preparation.
10. Preparation of Karyotypes 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, 2020, 2021 and 2022

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

Credits: 04 **MAXIMUM MARKS : 100**

Time Duration: 2 Hrs & 30 mins

a. Minor Test I : 20

b.Minor Test II : 20

c. Major Test : 60

Course Outcomes (CO)

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

CO-2: The students will get knowledge about Protein and Nucleic acid metabolism. Any change in biochemical pathways 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 genetic basis of reproductive disorders.

Unit–I

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

1.2 Disorders of Carbohydrates 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 & 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)

C.No. PSHGTC-201, Biochemistry of Metabolic Disorders & Developmental genetics (May2020-2022)

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. Developmental gene families
4.2. Limb as a development model
4.3. Role of Development genes in cancer
4.4. Sexual determination and differentiation
4.5. Hydatiform moles

Unit-V

5.1. Male and Female reproductive systems **(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 covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	20
Minor Test-II	21-40%	1 Hr	20
Major Test	41-100%	2 Hrs.& 30 mins	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. 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, 2020, 2021 and 2022

Course No. PSHGTC- 202 **Course Title: - Clinical Microbiology & Immunology**

Credits: 04 **MAXIMUM MARKS : 100**

Time Duration: 2 Hrs & 30 mins **a. Minor Test I : 20**

b. Minor Test II : 20

c. Major Test : 60

Course Outcomes (CO): -

CO-1: Course has been designed to make the student of Human Genetics Familiar with the Microbiology world, the structure, shape, nutrition & genetics of microbes, as they make the backbone of Genetic Engineering and Biotechnology.

CO-2: The students would be briefed about the pathogenic nature of the microbes which leads to many pathological complications in human.

CO-3: The course offers basics of immunology.

CO-4: Students will get complete understanding about activation & differentiation of immune cells. Further, the course will also help the students in learning the basics of MHC molecule and their role in human diseases.

CO-5: The students will be able to understand role of immune system in human health.

Unit-1

Introduction to Microbiology **(13 hrs.)**

1.1. History and Scope of Microbiology

1.2. Structure and organization of Microbial cells.

1.2.1 Structure of bacterial cell

1.2.1 Shape and type of bacteria

1.2.2 1.2.3 Introduction to viruses: their shapes and type

1.3 Microbial nutrition (Auxotrophs, Heterotrophs)

1.3.1 Aerobic and Anaerobic growth

1.3.2 Microbial Growth & growth curves

1.3.3 Toxins : Exotoxins & Endotoxins: Cholera toxin, Botox and Tetanus

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

Microbial diseases - Etiology, Pathogenesis and control of:

2.1 Air borne bacterial diseases with special reference to, Tuberculosis, Pneumonia, Diphtheria.

2.2 Water borne bacterial infection with special reference to Cholera, bacterial dysentery and Diarrohea, Salmonella infection and food poisoning.

2.3 Viral diseases:

2.3.1 AIDS

2.3.2 Hepatitis

2.3.3. HIN1 infection

Unit-III **(13 hrs.)**

Immunology-I

3.1 Introduction to Immune system

3.1.1 Innate & Acquired Immunity

3.1.2 Cells & organs of Immune system

3.1.3 Immune Response

3.2 Antigens: Immunogenicity vs Antigenicity

3.3 Structure of T & B cells

3.4 Immunoglobulins: Types, Structure & Function
3.5. Cytokines, their role in human diseases

Unit-IV **(13 hrs.)**
Immunology-II
4.1 Major Histocompatibility complex
4.2 Role of HLA in disease susceptibility
4.3 Transplantation immunology (Allograft, Xeograft, Syngraft, graft, graft rejection)
4.4 Antigen processing & presentation
4.5 T-cell maturation, activation and differentiation
4.6 B-cell maturation, activation and differentiation

Unit-V **(12 hrs.)**
Immune System in Human Health
5.1 Complement system
5.2 Hypersensitivity
5.3 Monoclonal antibodies: Production and Applications
5.4 Vaccines: Types and mode of action
5.5 Concept of Immunotherapy

Note for paper setting:

Examination theory weightage	Syllabus to be covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	20
Minor Test-II	21-40%	1 Hr	20
Major Test	41-100%	2 Hrs.& 30 mins	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. 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, Janis Kuby, 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 Higher Education, 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, 2020, 2021 and 2022

Course No. PSHGTC- 203 | **Course Title: - Human Molecular Genetics-II**

Credits: 04 | **MAXIMUM MARKS : 100**

Time Duration: 2 Hrs & 30 mins

a. Minor Test I : 20

b. Minor Test II : 20

c. Major Test : 60

Course outcome (CO)

CO-1: - Objectives of this course is to understand the principle & process of DNA cloning.

CO-2: The course will help the students to gather knowledge about pathology of different genetic diseases at molecular level.

CO-3: The course will emphasize to collect acquaintance on different strategies of diagnosis and testing individuals for genetic abnormalities.

CO-4: The objective of the course is to provide complete information about DNA sequencing, Transcirptome & protein profiling.

CO-5: Course has been designed to make the student of Human Genetics Familiar with different DNA diagnostic approaches.

Unit–I

DNA Cloning **(12 hrs.)**

2.1 Cell -based DNA cloning
- 2.1.1 Principles of DNA cloning
- 2.1.2 Cloning vectors: Plasmids, Phages, Cosmids,YAC

2.2 Expression cloning

2.3 Cell free DNA cloning
- 2.2.1 Principles of PCR
- 2.2.2 PCR types and its applications

2.4 DNA Fingerprinting

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

Genetic Testing in Individuals and Populations **(12 hrs.)**

3.1. Introduction.
3.2. Population Screening (Prenatal Screening, Newborn Screening, Carrier Screening)
3.3. Choice of material to test: DNA, RNA or Protein.
3.4. Scanning a gene for mutation.
3.5. Testing for a specified sequence change

Unit-IV

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

4.1. Methodology of DNA sequencing
4.2. Human Genome Project

4.3. Studying the Transcriptome.
- 4.3.1. Studying Transcriptome by sequence analysis.
- 4.3.2. Studying Transcriptome by Microarray or Chip analysis.

4.4 Studying the Proteome
- 4.4.1 Protein Profiling (2D Eclectrophoresis & MALDI-TOF)
- 4.4.2 Identifying Proteins that interact with one another (Yeast hybrid system, phage display
- 4.4.3 Protein degradation.

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

DNA Diagnostics

5.1. DNA Diagnostic approaches.

5.2. Etiology & Diagnosis of some common human Genetic Diseases:
- 5.2.1 Huntington chorea
- 5.2.2 Hemophilia
- 5.2.3 Thalassemia
- 5.2.4 Sickle Cell Anemia
- 5.2.5 Fragile-X syndrome
- 5.2.6 Cystic Fibrosis

Note for paper setting:

Examination theory weightage	Syllabus to be covered in examination	Time allotted	% marks
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Minor Test-II	21-40%	1 Hr	20
Major Test	41-100%	2 Hrs.& 30 mins	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. F Vogel A.G. Motulusky. Human Genetics: Problems and Approaches. 4th Edition, Springer-Verlag, 2010.
2. Tom Strachen , Human Molecular Genetics, 4th Edition, Garland Science, 2010.
3. D. Peter Snustad and Michael J.Simmons. Principles of Genetics. 6th edition. John Wiley & Sons, Inc., 2011.
4. Tom Strachen , Human Molecular Genetics, 5th Edition, Garland Science, 2014.
5. Robert L. Nussbaum, Roderick R. McInnes, & Huntington F. Willard, Thompson & Thompson Genetics in Medicine;, 8th edition, Imprint : Saunders, 2015.
6. Drs. Peter Turnpenny and Sian Ellard., Emery's Elements of Medical Genetics, 15th edition , Elsevier, 2017.
7. Terry A. Brown, Geneomes 4, 4th edition, Garland Science, 2017.
8. Lewin, Gene XII, 12th Edition, 2017.

M.Sc. Human Genetics

2nd Semester (CBCS)

Syllabus for the examination to be held in May, 2020, 2021 and 2022

Course No. PSHGTC-204 | **Course Title: - Human Cytogenetics**
Credits: 04 | **MAXIMUM MARKS : 100**
Time Duration: 2 Hrs & 30 mins | **a. Minor Test I : 20**
b. Minor Test II : 20
c. Major Test : 60

Course Outcomes

CO-1: The course has been designed with the objective to make the students of Human Genetics to learn about Human congenital defects and anomalies.
CO-2: Students will get knowledge about genetic basis of cancer.
CO-3: The course will offer provide knowledge about role of eugenics in society.
CO-4: Students will be able to have comprehensive understanding regarding genetic basis of autoimmune diseases and immnodefeciencies.
CO-5: The will focus on detecting genetic abnormalities by using advanced cytogenetic techniques.

Unit – I

Human Congenital Anomalities **(13 hrs.)**

1.1 Introduction.
1.2 Neural Tube Defects.
- 1.2.1 Anencephaly
- 1.2.2 Encephalocele
- 1.2.3 Hydrencephaly
- 1.2.4 Spina bifida including myelomeningocele and others

1.3 Cleft Lip/Cleft Palate.
1.4 Genomic Imprinting/ Uniparental Disomy
- 1.4.1 Prader-Willi Syndrome
- 1.4.2 Angelman Syndrome
- 1.4.3 Beckman Weidworth Syndrome

Unit-II

Cancers **(13 hrs.)**

2.1 Genetic basis of Cancers: Hallmark of Cancer cells
2.2 Environmental factors inducing cancers
2.3 Tumor Progression: Angiogenesis and Metastasis
2.4 Introduction to Proto-oncogenes, Oncogenes and Tumor suppressor genes
2.5 Role of Oncogenes in Cancer development (Burkitt's Lymphoma & Philadelphia chromosome).
2.6. Involvement of TSGs in;
- 1.7.1 Retinoblastoma
- 1.7.2. Breast cancer

Unit-III

Chromosome Instability Syndrome and Eugenics **(13 hrs.)**

3.1. Chromosome Instability syndrome: Ataxia Telangiectasia, Fanconi Anaemia.
3.2. Effect of mutagenic and teratogenic exposures in early pregnancy
3.3. Concept of artificial chromosome and its applications.
3.4. Eugenics
- 3.4.1. Definition
- 3.4.2. History
- 3.4.3. Positive and Negative Eugenics

3.5. Euphenics and Euthenics, Euthanasia

M.Sc. Human Genetics 2nd Semester (CBCS).
C. No. PSHGTC-204, Human Cytogenetics (May, 2020-22)

Unit-IV
Autoimmunity **(12 hrs.)**

4.1. Genetic Basis of Autoimmune Diseases
  4.1.1 Rheumatoid Arthritis
  4.1.2. Graves Disease
  4.1.3. Treatment of Autoimmune diseases
4.2. Immunodeficiencies
  4.2.1. Introduction
  4.2.2. Primary Immunodeficiency - SCID.
  4.2.3 Secondary Immunodeficiency – AIDS, Leukemia

Unit-V
Advanced Cytogenetics **(12 hrs.)**

5.1. Cytogenetics in Medicine
5.2. Analysis of Mitotic Chromosomes
5.3. FISH and its clinical application
5.4. Comparative Genomic Hybridization
5.5. Spectral Karyotyping

Note for paper setting:

Examination theory weightage	Syllabus to be covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	20
Minor Test-II	21-40%	1 Hr	20
Major Test	41-100%	2 Hrs.& 30 mins	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. 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 in Medicine;, 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)

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

Syllabus for the examination to be held in May, 2020, 2021 and 2022.

Note: Alterations will be done in the following listed practicals depending on the research work going on in the Major Research Lab of IHG with authorization of the Coordinator.

LAB COURSE NO. PSHGPC-205

List of practicals based on Theory Course no. 201 & 202

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-autoanalyzer
    ii. Fully autoanalyzer

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 autoanalyzer.
8. To perform lipid profiling of a given serum/blood sample on autoanalyzer.
9. To perform renal profiling of a given serum/blood sample on autoanalyzer.
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, Ambiguous genitalia).

M.Sc. Human Genetics

2nd Semester (CBCS)

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

Syllabus for the examination to be held in May, 2020, 2021 and 2022

Note: Alterations will be done in the following listed practicals depending on the research work going on in the Major Research Lab of IHG with authorization of the Coordinator.

LAB COURSE NO. PSHGPC-206

List of practicals based on Theory Course no. 203 & 204

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, 2020, 2021 and 2022

Course No. PSHGTC-301 | **Course Title: - Applied Medical Genetics**
Credits: 04 | **MAXIMUM MARKS : 100**
Time Duration: 2 Hrs & 30 mins | **a. Minor Test I : 20**
b. Minor Test II : 20
c. 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: Understanding the environmental, chromosomal and molecular etiology of Cancer.

CO-4: Course will help the students to have knowledge about treatment and management of genetic diseases.

CO-5: Students will come to gather information on genetic evaluation and treatment of human infertility.

Unit-I

Medical Genetics **(12 hrs.)**

1.1. History and impact of genetics in medicine
1.2. Clinical aspects of medical genetics
1.3. Animal models for the study of human genetic diseases
    1.3.1 Drosophila
    1.3.2 Yeast
    1.3.3 Mouse
1.4. Human mitochondrial DNA and related diseases

Unit-II

Studying Inheritance patterns **(13 hrs.)**

2.1. Inheritance pattern of genetic diseases
    2.1.1. Autosomal dominant disorders: Huntington Disorder
    2.1.2. Autosomal dominant disorders: Thalasemia and Sickle cell anemia
2.2. X-linked dominant disorders
        2.1.2.1. Familial rickets
        2.1.2.2. Hereditary nephritis
2.3. X- linked recessive disorders
    2.1.3.1. Color blindness
    2.1.3.2. Muscular dystrophies- BMD & DMD
2.4. Hemoglobin and Hemoglobinopathies
    2.4.1. Structure of hemoglobin
    2.4.2. Genetic control of hemoglobin synthesis
    2.4.3. Developmental control of globin gene
    2.4.4. Gene mutation and related abnormalities of hemoglobin

Unit-III

Chromosomal abnormalities in human cancers **(13 hrs.)**

3.1. Role of environment in carcinogenesis
3.2 Chromosomal changes associated with leukemias
3.3 Chromosomal changes associated with solid tumors
3.4 Chromosomal associated with benign tumors
3.5 Viral Oncogenesis
3.6 Association of HPV with human cervical carcinoma

M.Sc. Human Genetics
3rd Semester (CBCS)
C.No. PSHGTC-301, Applied Medical Genetics (Dec., 2020-2022)

Unit-IV

Managing Genetic Diseases

4.1. Prenatal testing
4.2. Pre-implantation of genetic diagnosis
4.3. Detection of genetic diseases
4.4. Treatment of genetic diseases
4.5. Management of genetic diseases
4.6. Consanguinity and its Consequences

Unit-V

Genetic Basis of Infertility

5.1. Cytogenetics 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

Note for paper setting:

Examination theory weightage	Syllabus to be covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	20
Minor Test-II	21-40%	1 Hr	20
Major Test	41-100%	2 Hrs.& 30 mins	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. 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, 2017.

M.Sc. Human Genetics
3rd Semester (CBCS)
Syllabus for the examination to be held in December, 2020, 2021 and 2022

Course No. PSHGTC-302 **Course Title: - Medical Biotechnology**
Credits: 04 **MAXIMUM MARKS : 100**
Time Duration: 2 Hrs & 30 mins **a. Minor Test I : 20**
b. Minor Test II : 20
c. Major Test : 60

Course outcomes (CO)

CO-1: The course will provide an opportunity to the students to have an exposure to the research methodologies being applied for the diagnosis of the human genetic diseases.
CO-2: Course has been designed to impart knowledge of gene therapy and its role in therapeutics to the students.
CO-3: Demonstration of stem cells relevance in cell based therapies will be given to the students.
CO-4: The course design will be focused on application of medical biotechnology in synthesis of different vaccines and peptide based drugs
CO-5: Providing knowledge to the students about pharmocogenomics and nanotechnology and how they will help to develop targeted drugs that will serve as a boon to the cancerous patients will be the prime motive of this course design.

Unit-I

Diagnostics **(12 hrs.)**

1.1. DNA diagnostics
1.2. Biochemical diagnostics
1.3. Immunodiagnostics
1.4. Prenatal diagnostics
1.4.1 Invasive techniques- Amniocentesis, Fetoscopy, Chorionic Villi Sampling
1.4.2. Non- invasive techniques- Ultrasonography, maternal fetal serum and fetal cells in the maternal blood.

Unit-II

Therapeutics **(13 hrs.)**

2.1. Concept of Gene therapy (ex-vivo and in-vivo approach)
2.2. Vectors used in gene therapy
2.2.1. Biological vectors- retrovirus, adenovirus, herpes
2.2.2. Synthetic vectors- liposomes, TFO, antisense therapy, Ribozymes, Protein aptamers,
2.3. Gene editing (CRISPR)
2.4. Strategies of gene therapy - Familial hypercholesterolemia, cystic fibrosis, ADA deficiency

Unit-III

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

3.1.Embryonic and adult stem cells
3.2. Characteristics of stem cell: Totipotent cells, Pluripotent cells, Multipotent cells
3.3. Culture of Stem cells
3.4. Human cord blood stem cells
3.5. Potential use of stem cells- cell based therapies
3.5.1. Current treatments
3.5.2. Potential treatments

Unit-IV

Applied Medical Biotechnology **(12 hrs.)**

4.1. Gene products in medicine
4.1.1. Anti- hemophilic factor
4.1.2. Humulin

4.1.3. Erythpoietin
4.1.4. Growth hormone/ somatostatin
4.1.5. Interferon
4.2. DNA based vaccines
4.2.1. Subunit vaccines- hepatitis B vaccine
4.2.2. Attenuated vaccines
4.3. Peptide based drugs

Unit-V

Medicine and the future **(13hrs.)**

5.1. Pharmacogenetics and Pharmacogenomics
5.2. Nanotechnology
5.2.1. Nanoparticles
5.2.2. Applications of Nanoparticles in medicine: Nanomedicine & Nanorobots
5.3 Nanotechnology in cancers
5.4. Overview of Genotoxicity

Note for paper setting:

Examination theory weightage	Syllabus to be covetred in examination	Time allotted	% marks
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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 Therapeutic Implementation, 2nd Edition, Academic Press, 2018.

M.Sc. Human Genetics
3rd Semester (CBCS)
Syllabus for the examination to be held in December, 2020, 2021 and 2022

Course No. PSHGTC- 303 **Course Title: - Population genetics, Bioinformatics & Biostatistics**

Credits: 04 **MAXIMUM MARKS : 100**

Time Duration: 2 Hrs & 30 mins

a. Minor Test I : 20

b. Minor Test II : 20

c. Major Test : 60

Course outcomes:

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

CO-2: This core course will be focused on the allelic frequencies calculation in population genetics.

CO-3: Behavioral genetics knowledge will be provided to the students in this course.

CO-4: A complete understanding on the use of bioinformatics in Human Genetics.

CO-5: The students will get knowledge of different biostatical tools used in analysis of genetic and non-genetic data.

Unit-I

Evolution **(12 hrs.)**

1.1. Origin of life
1.2 Concept and Theories of Evolution
1.3 Natural selection
1.4 Species and Speciation
1.5 Molecular Phylogeny
    1.5.1 Phylogenetic trees
    1.5.2 Construction of Phylogenetic trees
1.6. Evolution of Man

Unit-II

Population Genetics **(12 hrs.)**

2.1 Drawing and interpreting pedigrees
2.2 Calculations of Allelic frequencies
2.3 Hardy Weinberg Law
2.4 Consanguinity, Inbreeding & Inbreeding depression
2.5 Genetic drift: Bottle neck effect & Founders effect
2.6 Genetic Polymorphism

Unit-III

Behavior genetics **(12 hrs.)**

3.1 Personality disorders: Introduction
    3.2.1 Split Personality disorder
    3.2.2 Anxiety and Depression
3.2. Behavioral Genetics
    3.2.1. Schizophrenia
    3.2.2. Bipolar disorders
    3.2.3. Alcoholism

Unit–IV

Bioinformatics **(13 hrs.)**

4.1 Introduction
    4.1.1 Historical overview and definition
    4.1.2 Applications
4.2 Major databases in bioinformatics

4.2.1 Nucleic acid databases
4.2.2 Genome databases
4.2.3 Protein databases
4.3 Molecular biology and bioinformatics
4.4 Bioinformatics softwares
4.5 Information Search and Data Retrieval
4.5.1 The world wide web
4.5.2 Tools for web search
4.5.3 Data Retrieval tools

Unit- V **(13 hrs.)**

Biostatistics

5.1. Statistical Methods: Collection of data, Tabulation of data, Grouped and Ungrouped data, measures of central tendency and measures of dispersion, random experiment, measures of skewness and kurtosis, probability, Axiomatic definition, sample space, events.

5.2. Random variable : Discrete and Continuous Random variable. Poisson Distribution and give its Mean & Variance. Normal distribution and its characteristics.

5.3. Hypothesis and Testing : Statistic and Parameters, Population and sample size, Null and alternative hypothesis, Testing of Significance Tests (Z-test, F-test and Chi Square test), Odds Ratio.

Note for paper setting:

Examination theory weightage	Syllabus to be covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	20
Minor Test-II	21-40%	1 Hr	20
Major Test	41-100%	2 Hrs.& 30 mins	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. 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, 2020, 2021 and 2022

Course No. PSHGTO-304 **Course Title: - Human Genetic Disorders & Society-I**
Credits: 04 **MAXIMUM MARKS : 100**
Time Duration: 2 Hrs & 30 mins **a. Minor Test I : 20**
b. Minor Test II : 20
c. Major Test : 60

Course Outcomes:
CO-1: The course is designed with the objective to provide sound knowledge to the students about the basics of chromosomes and the various techniques used for chromosome analysis.
CO-2: Students will be briefed about the different disorders of autosomes and sex chromosomes.
CO-3: A detailed mechanism of DNA replication and its expression will be described to the students during this course study.
CO-4: Students will be made aware about the genetic aspects of different types of cancer and how they can be prevented by modifying their life style.
CO-5: A comprehensive knowledge about the complexity of multifactorial disorders will be provided to the students.

Unit-I
Chromosomes and cell division **(12 hrs.)**
1.1 Structure and functions of chromosomes
1.2 Human chromosomes
1.3 Cell division:
1.3.1 Mitosis
1.3.2 Meiosis
1.4 Analysis of human chromosomes
1.4.1. Conventional techniques
1.4.2. Advanced techniques

Unit-II
Disorders of autosomes and sex chromosomes **(12 hrs.)**
2.1 Disorders of the autosomes
2.2.1 Down syndrome
2.2.2 Edward syndrome
2.2.3 Patau syndrome
2.2 disorders of the sex chromosomes
2.3.1 Turner syndrome
2.3.2 Klinefelter syndrome
2.3 Sex limited, sex linked and sex influenced traits

Unit-III
Central Dogma of Life **(11hrs.)**
3.1 DNA as a genetic material
3.2 DNA Replication
3.3 Transcription
3.4 Translation

Unit-IV **(12 hrs.)**
Genetics and Cancer
4.1. Genetic and environmental factors in cancers
4.2. Genetics of common cancers
4.2.1. Leukemias

4.2.2. Breast cancer
4.2.3. Cervical cancer
4.2.4. Ovarian cancer

Unit-V **(13 hrs.)**

Genetics of Multifactorial diseases

5.1. Diabetes mellitus
5.2. Cardiovascular diseases
5.3. Hyperthyroidism
5.4. Schizophrenia
5.5. Parkinson's Disease

Note for paper setting:

Examination theory weightage	Syllabus to be covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	20
Minor Test-II	21%1-40%	1 Hr	20
Major Test	41%-100%	2 Hrs.& 30 mins	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. D. Peter Snustad and Michael J.Simmons. Principles of Genetics. 6th edition. John Wiley & Sons, Inc., 2011.
3. Robert Nussbaum et al. Thompson & Thompson genetics in Medicine, 8th Edition, Elsevier, 2015.
4. Helen M Kingston ABC of Clinical genetics, 4th Edition, BMJ, 2015.
5. Drs. Peter Turnpenny and Sian Ellard., Emery's Elements of Medical Genetics, 15th edition , Elsevier, 2017.
6. Emerys & Rimoin, Principles & Practice of Medical Genetics, 7th Edition, Elsevier, 2017.

M.Sc. Human Genetics
3rd Semester (CBCS)

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

Syllabus for the examination to be held in December, 2020, 2021 and 2022

Note: Alterations will be done in the following listed practicals depending on the research work going on in the Major Research Lab of IHG with authorization of the coordinator.

LAB COURSE NO. PSHGPC-305

List of practicals based on Theory Course no. 301 & 302

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 severity of 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 chromosomes with 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 karyotypes of the Infertile couples.

LAB COURSE NO. PSHGPC-306

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

List of practicals based on Theory Course no. 303

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,
IVth Semester (CBCS)
Syllabus for the examination to be held in May, 2021, 2022 and 2023

Course No. PSHGTC-401 | **Course Title: - Genetic Counselling**
Credits: 04 | **MAXIMUM MARKS : 100**
Time Duration: 2 Hrs & 30 mins | **a. Minor Test I : 20**
b. Minor Test II : 20
c. Major Test : 60

Course Outcomes

CO-1: The course has been designed with the objective to make students learn about the importance of Genetic Counselling 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 in detecting different 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 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 on Pre-conception and Pre-natal Diagnosis Act in Human Genetics.

Unit-I

Genetic counselling **(12 hrs.)**

1.1 Genetic counseling: an introduction
1.2 Genetic counseling in Mendelian disorders
1.3 Genetic counseling in common non- Mendelian disorders
1.4 Genetic counselors as educators
1.5 Risk assessment as a part of Genetic counseling

Unit-II

Genetic counselling and genetic disorders **(13 hrs.)**

2.1 Neuromuscular diseases
2.2 Central nervous system orders
2.3 Disorders of mental functions
2.4 Disorders of bone and connective tissues
2.5 Oral and craniofacial disorders
2.6 Deafness and renal diseases

Unit-III

Issues in Genetic testing-I **(12 hrs.)**

3.1. Genetic testing issues
    3.1.1. Privacy and Confidentiality
    3.1.2. Genetic Discrimination
3.2. Genetic Counselling registers
3.3 Genetic counselling clinics and its working
3.4 Objectives and Outcomes of Genetic Counselling

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 Counsellers

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 counselling 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 covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	20
Minor Test-II	21%-40%	1 Hr	20
Major Test	41%-100%	2 Hrs.& 30 mins	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
IVth Semester (CBCS)
Syllabus for the examination to be held in May, 2021, 2022 and 2023

Course No. PSHGTC-402 | **Course Title: - Clinical Genetics**
Credits: 04 | **MAXIMUM MARKS : 100**
Time Duration: 2 Hrs & 30 mins | **a. Minor Test I : 20**
b. Minor Test II : 20
c. 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

1.1 Principles and practice of clinical genetics **(12 hrs.)**
1.2 Molecular and Biochemical basis of genetic diseases
1.3 Late onset genetic disorders- Alzheimer, Parkinson's Disease
1.4 Nature and Nurture: Distangling the effects of genes and environment

Unit-II

2.1 Cytogenetic techniques in disease detection **(12 hrs.)**
2.2. Chromosome abnormalities and pregnancy loss
2.3. Ring chromosome and related genetic disorders
2.4. Chromosomal rearrangements and their impact on human health
2.5. Reprogenetics- Germinal Choice Technology

Unit-III

3.1. Treatment of genetic diseases **(13hrs.)**
- 3.1.1. Conventional approaches to treatment of genetic disease
- 3.1.2. Therapeutic application of recombinant DNA technology

3.2. Genetic susceptibility
3.3. Neonatal screening
3.4. Genetic registers
3.5. Fetal treatment

Unit – IV **(12 hrs)**

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
- 4.2.5 Schizophrenia

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

M.Sc. Human Genetics, IVth Semester (CBCS)
C.No. PSHGTC-402, Clinical Genetics (May, 2021-2023)

Note for paper setting:

Examination theory weightage	Syllabus to be covetred in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	20
Minor Test-II	21-40%	1 Hr	20
Major Test	41-100%	2 Hrs.& 30 mins	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
IVth Semester (CBCS)

Course Title: - Human Genetic Disorders & Society-II **Course No. PSHGTO-403**
Credits: 04 **MAXIMUM MARKS : 100**
Time Duration: 2 Hrs & 30 mins **a. Minor Test I : 20**
b. Minor Test II : 20
c. Major Test : 60

Syllabus for the examination to be held in May, 2021, 2022 and 2023.

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 Counselling in the welfare of family as well as society.

Unit-I
Recombinant DNA Technology **(13 hrs.)**
1.1 Cloning vectors: Plasmids, Phages, Cosmids, YAC
1.2 Enzymes used in RDT
1.2.1 Restriction Endonucleases
1.2.2 Other enzymes
1.3 Gene cloning
1.4 Ethical issues in gene cloning
1.5 Applications of RDT in Human Diseases

Unit-II
Single Gene Disorders **(12 hrs.)**
2.1. Hemophilia
2.2 Cystic Fibrosis
2.3 Sickle cell anemia
2.4 Huntington disease
2.5 Fragile X Syndrome

Unit-III
Molecular Genetic techniques **(13 hrs.)**
3.1. PCR: principle, working and its applications.
3.2. Electrophoresis (Overview)
3.3. FISH
3.4. DNA fingerprinting
3.5. DNA Sequencing:
3.5.1 Chain termination method
3.5.2 Chemical degradation method
3.5.3 Automated DNA sequencing.2 Enzymes used in RDT

Unit-IV
Rare Genetic Diseases **(12hrs.)**
5.1. Introduction to rare genetic diseases
5.2. Lysosomal storage diseases
5.3. Progeria
5.4. Werewolf syndrome

Unit V
Genetic Counselling **(13 hrs.)**
5.1. Genetic counselling: an introduction
5.2 Pedigree analysis
5.3 Consanguinity and its impact on Human health

5.4 Issues in genetic testing
  5.4.1 Privacy & Confidentiality
  5.4.2 Genetic discrimination
5.5 Genetic counselling registers
5.6 Role of PC and PNDT Act, 1994 in genetics

Note for paper setting:

Examination theory weightage	Syllabus to be covered in examination	Time allotted	% marks
Minor Test-I	Upto 20%	1 Hr	20
Minor Test-II	21%-40%	1 Hr	20
Major Test	41-100%	2 Hrs.& 30 mins	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

Ist Semester (CBCS)

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

Syllabus for the examination to be held in May, 2021: May, 2022 and May, 2023

Note: Alterations will be done in the following listed practicals depending on the research work going on in the Major Research Lab of IHG with authorization of the Coordinator.

LAB COURSE NO. PSHGPC-405

List of practical's based on Theory Course no. 401 & 402

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 proformas 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 diseases.