Course No. PSHGTC-101 **Title: - Cell Biology, Research Methodology and Instrumentation**

Credits: 04
Duration of examination: 2 Hrs & 30 mins

Maximum Marks
a) Semester examination: 60
b) Sessional examination: 40
Minor 1=20
Minor-2=20 Total=40

CELL BIOLOGY , RESEARCH METHODOLOGY AND INSTRUMENTATION

Objective: - The course has been designed to expose the student of Human Genetics to the structure and function of cell and its organelles. Cell – cell interaction, Signal transduction and Programmed cell death are some of the mechanism which will make a student to have better understanding of the cell. The course also lays emphasis on understanding the principle and working of the instruments and various research techniques available for the diagnosis of different diseases/disorders.

UNIT-I

Cell, Plasma Membrane and Cytoskeleton

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-II

Structure and Functions of Cell Organelles

2.1. Mitochondria
2.2. Ribosome
2.3. Golgi Complex
2.4. Endoplasmic Reticulum
2.5. Peroxisomes and Lysosomes
2.6. Nucleus

UNIT-III

Cell Cycle and Cell Signalling

3.1. Cell cycle and its regulation
3.2. Cell-Cell Interaction
  3.2.1. Cell adhesion molecules
  3.2.2. Cellular Junctions
  3.2.3. Extracellular matrix
3.3. Signal transduction
  3.3.1. Intracellular receptor and cell surface receptors
  3.3.2. Signalling via G-protein linked receptors (PKA, PKC, CaM kinase)

3.3.3. Enzyme linked receptor signaling (Growth factor receptor signaling; JACK-STAT pathway)

3.3.4. Network and cross-talk between different signal mechanisms

3.4. Programmed cell death (Apoptosis)

UNIT-IV

4.1 Centrifugation:

Basic principle, Types and its applications.

4.2 Chromatography:

4.2.1 Principle, Types and applicatons

4.2.2 Paper Chromatography,

4.2.3 Thin layer Chromatography:

4.2.4 Column Chromatography:

4.2.4.1 Ion-exchange

4.2.4.2 Gel-filtration

4.2.4.3 HPLC

4.2.4.4 Affinity columns

4.3 Electrophoresis

Principle, Types and applications.

UNIT-V

5.1 Microscopy : Basic principles & applications

5.1.1 Light Microscopy

5.1.2 Dark-field Microscopy

5.1.3 Phase-contrast Microscopy

5.1.4 Fluorescence Microscopy

5.1.5 Electron Microscopy

5.2 Immunological Techniques

5.2.1 Immunodiffusion

5.2.2 Immunoelectrophoresis

5.2.3 Immunofluorescence

5.3 RIA, ELISA & Western Blotting

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

Major test:

i. Major test will have seven questions each of 15 marks

ii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus

iii. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

Books Recommended

1. Geoffrey M. Copper and Robert E. Hausman. 2009. The Cell : A Molecular Approach, Fifth Edition. ASM Press and Sinauer Associates, Inc.
2. Gerald Karp, 2009. Cell and Molecular Biology : Concepts and Experiments, 6th Edition John Wiley and Sons.
3. Bruce Alberts et.al, 2007. Molecular Biology of the Cell, 5th Edition, Taylor & Francis Group.
4. Alberts *et al, 2002.Molecular* Biology of the Cell
5. De-Robertis,2003. Cell & Molecular Biology (latest Edition)
6. Powar, C.B., 2000. Cell Biology (latest Edition)
7. Rastogi, V.B., 2007. Cell Biology. Third Edition. New Age International Publishers.

Course No. PSHGTC- 102

Title:- Human Anatomy,Embryology and Endocrinology

Credits: 04

Duration of examination: 2 Hrs & 30 mins

Maximum Marks:

a) Semester examination: 60

b) Sessional examination: 40

Minor 1=20

Minor-2=20 Total=40

HUMAN ANATOMY, EMBRYOLOGY AND ENDOCRINOLOGY

Objectives: - Course is designed to apprise the student about the details of different systems of human body and the process from fertilization to implantation of fertilized ovum. This will helps the students to understand the changes caused in different systems and organs due to genetic changes.

UNIT-I

1.1 Muscular & Skeletal system:

1.1.1. Classification of Bones
1.1.2. Ossification and growth of Bones
1.1.3. Joints and their types
1.1.4. Classification of muscles
1.1.5. Structure of smooth, cardiac and skeletal muscles
1.1.6. Neuromuscular junction
1.1.7. Degenerative disorders of muscles

UNIT-II

2.1. Digestive system:

2.1.1 Introduction
2.1.2 Physiology of Digestion

2.2 Circulatory system:

2.2.1. Blood and Lymph
2.2.2 Structure of Heart and its functioning

2.3. Respiratory system

2.3.1. Parts of Respiratory system
2.3.2 Physiology of Respiration

2.4. Excretory system:

2.4.1 Structure and function of Nephron
2.4.2 Physiology of Excretion

UNIT-III

3.1 Nervous system:

3.1.1 Structure of Brain & Spiral Cord

3.2 Sensory Organs

3.2.1 Eyes
3.2.2 Ears
3.2.3 Nose
3.2.4 Skin

UNIT-IV

4.1 Physiology of Endocrine System

4.1.1 Pituitary Gland
4.1.2 Thyroid, Parathyroid Gland
4.1.3 Adrenal Gland
4.1.4 Islets of Langerhans
4.1.5 Gonads
4.1.6 Pineal Gland
4.1.7 Thymus Gland

UNIT-V

5.1 Embryology

5.1.1 Fertilization & Implantation
5.1.2 Dev. of human embryo upto three germinal layers
5.1.3 Dev. of embryonic disc, notochord formation & neurulation
5.1.5 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

Major test:

i. Major test will have seven questions each of 15 marks
ii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus
iii. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

Books Recommended:-

1. Turner, S.L. (1966). General Endocrinology W.S. Saunders, Tappan Co.Ltd.Tokyo, Japan.
2. Guyton and Hall. Text book of Medical Physiology. 12th Edition. Elsevier Saunders Publishers
3. J.S.Lewis. The Endocrine System. (latest Edition)
4. J. Matthew Neal, 2001. How the Endocrine system works, Blackwell Science.
5. Roger Lewin. Hormones. (latest Edition)
6. Harper's 28th Edition, Illustrated Biochemistry. The McGraw- Hill Compa

Course No. PSHGTC- 103 **Title:- Human Molecular Genetics-I**
Credits: 04

Duration of examination: 2 Hrs & 30 mins **Maximum Marks:**

c) Semester examination: 60

d) Sessional examination: 40

Minor 1=20

Minor-2=20 Total=40

HUMAN MOLECULAR GENETICS-I

Objective: - Human Molecular Genetics is a vast field that provides information of Genetic Material, general principles and applications of cloning and molecular hybridization. It provides comprehensive guide to the structure, function and evolution of the human genome and human genes.

UNIT-I

Nucleic Acid : Structure & functions

1.1 DNA – Structure, types & functions
1.2 DNA Replication in Prokaryotes & Eukaryotes.
1.3 RNA - Structure, types & functions
1.4 Transcription factors & Gene Expression
1.5 Post transcripted RNA processing
1.6 Translation : Protein formation

UNIT-II

2.1 Cell -based DNA cloning
- 2.1.1 Principles of DNA cloning
- 2.1.2 Vector Systems for cloning different sizes of DNA fragments
- 2.1.3 Expression cloning

2.2 PCR based DNA cloning & DNA Analysis
- 2.2.1 Principles of PCR
- 2.2.2 Applications of PCR
- 2.2.3 Real Time PCR

UNIT-III

Human Genome

3.1 Basic concepts of Human Genome
3.2 Human Gene Families
3.3 Homology, Parelogs & Orthologs
3.4 Mitochondrial Genome
3.5 Repetitive DNA and its types
3.6 Transposable elements in Eukaryotes**.**

UNIT-IV

4.1 DNA hybridization assays
- 4.1.1 Nucleic acid probes
- 4.1.2 Principles of molecular hybridization
- 4.1.3 Methods and applications of molecular hybridization.

4.1.4 Synthesis and labeling of probes

UNIT-V

5.1 Molecular evolution

5.1.1 Evolution of human mitochondrial genome.
5.1.2 Evolution of human nuclear genome.
5.1.3 Evolution of human sex chromosomes.
5.1.4 Retroposons

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

Major test:

i. Major test will have seven questions each of 15 marks

ii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus

iii. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

BOOKS RECOMMENDED

1) F Vogel A.G. Motulusky. Human Genetics: Problems and Approaches. Third Completely Revised Edition, Springer-Verlag.
2) D. Peter Snustad and Michael J.Simmons. Principles of Human Genetics. Fifth edition. John Wiley & Sons, Inc.
3) Ricki Lewis. 2009 Human Genetics-Concepts and Application. Second Edition. WCB-McGraw Hill.
4) Benjawin Lewin, 2008. Gene IX. Jones and Barlett Publishers.
5) T.A.Brown, 2002.Genome,Second Edition, Bios Scientific Publishers Ltd.
6) T. A. Brown, (2006): Genome : Third Edition, Garland Science.
7) David P. Clark, 2005. Molecular Biology. Elsevier Academic Press.

Course No. PSHGTC- 104 **Title: - Human Cytogenetics - I**

Credits: 04
Duration of examination: 2 Hrs & 30 mins **Maximum Marks:**

e) Semester examination: 60
f) Sessional examination: 40
Minor 1=20
Minor-2=20 Total=40

HUMAN CYTOGENETICS-I

Objectives: The course has been designed to provide an introduction to Human Cytogenetics. Different steps and advances that occurred in understanding human chromosomes have been discussed in detail. Structural details and the role of chromosomes in human congenital anomalies & cancers have been discussed so that a student pursuing P.G course in Human Genetics is able to understand the importance of human chromosomes.

UNIT-I

1.1 Introduction to Human Cytogenetics & Human Chromosomes.
1.2 Mendel's Laws of Heredity.
1.3 Chromosomal Theory of Heredity / Inheritance and Non disjunction as a proof to Chromosomal Theory
1.4 The Human Karyotype

UNIT-II

2.1 Chromosome structure and Organization.
2.2 Types of cell division – Mitosis and Meiosis
2.3 Techniques for Chromosomes study
- i. G-Banding
- ii. C-Banding
- iii. Q-Banding
- iv. R-Banding
- v. High Resolution Banding

2.4 Tissue culture techniques
- i. Whole Blood Culture
- ii. Bone Marrow Culture
- iii. Aminocyte Culture
- iv. Skin Fibroblast

UNIT-III

3.1 Numerical Changes in Autosomes & Sex Chromosomes.
3.2 Structural Changes in Autosomes & Sex Chromosomes – Chromosomes breaks & rearrangements.
3.3 Telomeres – Structure & Capping
3.4 Chromosome Abnormalities in Human Pregnancies.

UNIT-IV

4.1. Mutations
- 4.1.1 Germ line & Somatic Mutations
- 4.2.2 Physical & Chemical Mutagens

4.2 Molecular Basis of mutations
- 4.2.1 Nucleotide Subotitutions
- 4.2.2 Mis sense mutations.
- 4.2.3 Insertions, Deletions & Frameshift Mutations.
- 4.2.4 Dynamic Mutations of Trinucleotide Repeats.

UNIT-V

5.1. Inactivation of Sex Chromosomes

5.2. Human Y-Chromosome
- 5.2.1. Structure
- 5.2.2. Genes on Y
- 5.2.3. X & Y pairing & Pseudoautosomal Region
- 5.2.4. Sex determination in Human

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

Major test:

i. Major test will have seven questions each of 15 marks

ii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus

iii. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

Books Recommended:

1. Emery and Rimoin's, et al., 2007. Principles and Practice of Medical Genetics. Fifth Edition. Volume – II, Churchill Livingstone Elsevier.
2. Emery and Rimoin's, Principles and Practice of Medical Genetics. 3rd Edition. – 3 Volume Set, Churchill Livingstone Elsevier.
3. De Grouchy & Turleau.1984. Clinical atlas on Human Chromosomes.
4. Jankowski & Polak, 1996.Clinical Gene Analysis and Manipulation.
5. Robinson and Linden, Clinical Genetics Handbook. (latest Edition)
6. F Vogel A.G. Motulusky. Human Genetics: Problems and Approaches. Second Completely Revised Edition, Springer-Verlag. (latest Edition)
7. Golder N. Wilson, M.D., Ph.D.Clinical Genetics-A Short Course. A John Wiley and Sons, Inc., Publication. (latest Edition)
8. G.S. Miglani – Fundamentals of Genetics, 2008
9. Darbeshwar Roy – Cytogenetics, 2009
10. Hartl & Jones – Genetics (Analysis of Genes & Genomes), Seventh Edition.

Course No. PSHGTC- 201 **Title: - Biochemistry of Metabolic Disorders & Biostatistics**
Credits: 04
Duration of examination: 2 Hrs & 30 mins **Maximum Marks;**

c) **Semester examination: 60**
d) **Sessional examination: 40**
Minor 1=20
Minor-2=20 **Total=40**

BIOCHEMISTRY OF METABOLIC DISORDERS & BIOSTATISTICS

Objectives: - The course has been designed to provide the student details about the Biochemical pathways in human body. 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**.**

Unit–I

1.1 Introduction to Carbohydrates Metabolism
1.2 Disorders of Carbohydrates Metabolism
- 1.2.1 Lactose Intolerance
- 1.2.2 Glycogen Storage Disease (G-6PD)
- 1.2.3 Fructose Intolerance
- 1.2.4 Diabetes Mellitus
- 1.2.5 Galactosemia

Unit–II (Unit- completed)

2.1 Introduction to Proteins & Amino acids.
2.2 Disorders of Amino acids metabolism
- 2.2.1 Phenylketonuria
- 2.2.2 Alkaptonuria
- 2.2.3 Tyrosinemia

2.2.4 Albinism
2.3 Metabolic Disorders of Purines and Pyrimidines
- 2.3.1 Hyperuricemia
- 2.3.2 Lesch-Nyhan Syndrome

2.4 Metabolic disorders of Porphyrin
- 2.4.1 Acute Intermittent Porphyrin
- 2.4.2 Erythropoietic Porphyria

2.5 Metabolic disorders of Glycosamineglycans & Glycoproteins
- 2.5.1 Mucopolysaccharidosis
- 2.5.2 Mucolipidosis

Unit–III

3.1 Introduction to Lipids & Fatty acids & their metabolism
3.2 Disorders of Lipid Storage
- 3.2.1 Tay Sachs Disease
- 3.2.2 Krabbe Disease

3.3 Disorders of Fatty acid Metabolism
- 3.3.1 Hyperlipidemia
- 3.3.2 Hypercholesterolemia

Unit–IV

Bioinformatics (Guest faculty available)

4.1 Introduction
- Historical overview and definition
- Applications
- Major databases in bioinformatics

a) Nucleic acid databases
b) Genome databases
c) Protein databases

- Molecular biology and bioinformatics
- Bioinformatics softwares

4.2 Information Search and Data Retrieval

- The world wide web
- Tools for web search
- Data Retrieval tools

UNIT- V

Biostatistics

5.1 Statistical Methods: Collection of data, Tabulation of data, Grouped and Ungrouped data, Classes : their explanations, frequency distribution and its graphical representation Frequency tables, cumulative frequency, 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. Binomial Distribution & give its mean and variance. 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)

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

Major test:

iv. Major test will have seven questions each of 15 marks

v. One question will be very short answer type of multiple parts compulsory spread over entire syllabus

vi. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

BOOKS RECOMMENDED

1. Lubert Stryer's; Biochemistry, Fifth Edition, published by W.H. Freeman and Company.
2. Harpers illustrated Biochemistry (28th edition) The Mc Graw Hill Companies, 2009
3. Biochemistry by Donald Voet (3rd edition) pubisher : Wiley
4. Jeremy *et al, 2002.Biochemistry*
5. Lehninger Principles of Biochemistry (3rd Ed.), MacMillan Worth, 2000
6. Berg et al.: Biochemistry (5th Ed.), Freeman, 2002
7. Mathews et al.: Biochemistry (3rd Ed.), Pearson, 2004
8. Zubay et al: Principles in Biochemistry (2nd Ed.), WCB, 1995
9. Rawn: Biochemistry, Neil Patterson, 1989
10. Mahler & Cordes: Textbook of Biological Chemistry, Harper, 1966
11. Bioinformatics for Geneticists, Michael Barnes, Ian C Gray (Editors), 2003, John Wiley & Sons

12. Bioinformatics for Dummies, Jean-Michel Claverie, Cedric Notredame, 2003, John Wiley & Sons
13. Mathematics of Genome Analysis, Jerome K. Percus, 2002, Cambridge Univ Press
14. Bioinformatics Computing, Bryan P. Bergeron, 2002, Prentice Hall
15. Evolutionary Computation in Bioinformatics, Gary B. Fogel, David W. Corne (Editors), 2002, Morgan Kaufmann
16. Introduction to Bioinformatics, Arthur M. Lesk, 2002, Oxford University Press
17. Instant Notes in Bioinformatics, D.R. Westhead, J. H. Parish, R.M. Twyman, 2002, Bios Scientific Pub
18. Fundamental Concepts of Bioinformatics, Dan E. Krane, Michael L. Raymer, Michaeel L. Raymer, Elaine Nicpon Marieb, 2002, Benjamin/Cummings
19. Essentials of Genomics and Bioinformatics, C. W. Sensen (Editor), 2002, John Wiley & Sons.
20. Current Topics in Computational Molecular Biology (Computational Molecular Biology), Tao Jiang, Ying Xu, Michael Zhang (Editors), 2002, MIT Press.
21. Fundamental of Statistics by V.K. Kapoor and S.C. Gupta.
22. Statistical Methods by S.C. Gupta.

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

Credits: 04
Duration of examination: 2 Hrs & 30 mins

Maximum Marks;

a) Semester examination: 60
b) Sessional examination: 40
Minor 1=20
Minor-2=20 Total=40

CLINICAL MICROBIOLOGY AND IMMUNOLOGY

Objectives: - Course has been designed to make the student of Human Genetics Familiar with the Microbiology world as they make the backbone of Genetic Engineering and Biotechnology. An understanding the microbial cell helps in learning the basic processes of Physiology and Biochemistry with special reference to metabolic pathways.

UNIT-1

Introduction to Microbiology

1.1 History and Scope of Microbiology
1.2 Structure and organization of Microbial cells.

1.2.1 Structure of bacterial cell
1.2.2 Shape and type of bacteria
1.2.3 Introduction to viruses
1.2.4 Shape and type of viruses

UNIT-II

Physiology and Biochemistry of Microbes

2.1 Microbial nutrition
2.2 Aerobic and Anaerobic growth
2.3 Microbial Growth
  2.3.1 Growth Curve
  2.3.2 The Mathematics of growth
2.4 Toxins : Exotoxins & Endotoxins
2.5 Microbial degradation

UNIT-III

Microbial diseases - Etiology, Pathogenesis and control of :

3.1 Air borne bacterial diseases with special reference to, Tuberculosis, Whooping cough, Diphtheria, Streptococcal infection, Staphylococcal infection and Diplococcal infection.
3.2 Water borne bacterial infection with special reference to Cholera, bacterial dysentery and Diarrohea, Salmonella infection, and food poisoning.
3.3 Viral oncogenesis
3.4 AIDS
3.5 Hepatitis

UNIT-IV

Immunology

4.1 Introduction to immunity
  4.1.1 Active & Passive Immunity
  4.1.2 Innate & Acquired Immunity
  4.1.3 Immune System
4.2 Immune Response
4.3 Cells of Immune system & immune modulators
4.4 Structure of T & B cells: Antigen processing, presentation & cell proliferation.
4.5 Immunoglobulins.

UNIT-V

5.1 Immunogenetics & Vaccines

5.1 Monoclonal antibodies: Production and Applications
5.2 Cytokines, Interleukins and their regulation
5.3 Vaccines: Types and mode of action
5.4 Principles of vaccine preparation
5.5 Major Histocompatibility complex

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

Major test:

i. Major test will have seven questions each of 15 marks
ii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus

iii. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

BOOKS RECOMMENDED

1. Prescott, Harley, Klein; Microbiology, 6th edition, Mc Graw- Hill Higher Education
2. Pelczar, Michael J. Jr. / Chan, E.C.S / Krieg, Noel R., Microbiology, 37th reprint 2003, Mc Graw- Hill Higher Education
3. Schlegal, M.G., 1985. General Microbiology, Cambridge University Press.
4. Immunology by Richard A. Goldsby (Editor), Barbara A. Osborne, Thomas J. Kindt, Janis Kuby, Janis Kuby, Richard A. Goldby, 6th edition, whfreeman press co.
5. Clinical Immunology by Pravash Sen Gupta, Oxford University Press.
6. Roitt's Essential Immunology, 11th edition, Wiley-Blackwell Co.
7. Text book of immunology by Barret, Latest edition

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

Credits: 04

Duration of examination: 2 Hrs & 30 mins

Maximum Marks;

a) Semester examination: 60

b) Sessional examination: 40

Minor 1=20

Minor-2=20 Total=40

HUMAN MOLECULAR GENETICS -II

Objectives: - Objectives of this course is to understand the molecular basis of human genetics that has become essential in diagnosing and treating the human genetic diseases. Modern methods for detecting defective gene, and the basis of positional cloning to map the disease genes helps in counselling to the affected families.

Unit–I

Human Genetics Diseases

1.1. Principle and strategies in identifying human disease genes.
1.2. Position independent strategies for identifying disease gene.
1.3. Positional cloning.
1.4. Confirming a candidate gene.
1.5 Ways to identify disease genes.

Unit-II

Molecular Pathology

2.1. Introduction.
2.2. Rules for nomenclature of mutations and databases of mutation.
2.3. Loss of function mutations.
2.4. Gain of function mutations.
2.5. Molecular pathology: from gene to disease.
2.6. Molecular pathology: from disease to gene.
2.7. Molecular pathology of chromosomal disorders.

Unit – III (covered)

Genetic Testing in Individuals and Populations

3.1. Introduction.
3.2. Choice of material to test: DNA, RNA or Protein.
3.3. Scanning a gene for mutation.
3.4. Testing for a specified sequence change.
3.5. Gene tracking.
3.6. Population Screening.

UNIT-IV

Genome Sequence and Function

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
  4.4.2. Identifying Proteins that interact with one another.
  4.4.3. Protein degradation.

Unit-V

DNA Diagnostics

5.1. DNA Diagnostic approaches.
5.2. DNA based 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 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

Major test:

i. Major test will have seven questions each of 15 marks
ii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus
iii. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

BOOKS RECOMMENDED

1. Thompson & Thompson Genetics in Medicine; Robert L. Nussbaum, Roderick R. McInnes, & Huntington F. Willard, 7th edition, Imprint : Saunders.
2. Emery's Elements of Medical Genetics, 14th edition by Drs. Peter Turnpenny and Sian Ellard.
3. Terry A. Brown, Geneomes 3, 2006. 3rd edition. Garland Science.

4. F Vogel A.G. Motulusky. Human Genetics: Problems and Approaches. Second Completely Revised Edition, Springer-Verlag.
5. D. Peter Snustad and Michael J.Simmons. Principles of Human Genetics. Third edition. John Wiley & Sons, Inc.
6. Ricki Lewis. Human Genetics-Concepts and Application. Second Edition. WCB-McGraw Hill.
7. Lewin, 2004. Gene VIII
8. T.A.Brown.Genome,Second Edition.
9. Brown, T. A. (1989): Genetics : A Molecular Approach, VNR International
10. Peter Russel: I Genetics-A Molecular approach
11. Human Molecular Genetics: Tom Strachen Ed.II and III, Garland Science

Course No. PSHGTC- 204 **Title: - Human Cytogenetics- II**

Credits: 04
Duration of examination: 2 Hrs & 30 mins **Maximum Marks;**

a) Semester examination: 60

b) Sessional examination: 40
Minor 1=20
Minor-2=20 Total=40

HUMAN CYTOGENETICS - II

Objectives: The course has been designed with the objective to make the students of Human Genetics to learn about genetics of immediate importance to the society. Human Society is currently being confronted with problems arising from lack of knowledge about Human Genetics and application of this knowledge to the welfare of the society.

Unit – I

Cancers

1.1. Genetic basis of Cancers
1.2. Oncogenes & Proto-oncogenes
1.3. Tumor Suppressor Genes
1.4. Genetics of common Cancers

UNIT-II

Human Congenital Anomalities

2.1. Introduction.
2.2. Neural Tube Defects.
  2.2.1. Anencephaly
  2.2.2. Encephalocele
  2.2.3. Hydrencephaly
  2.2.4. Spina bifida including myelomeningocele and others
2.3. Cleft Lip/Cleft Palate.
2.4. Uniparental Disomy
  2.4.1. Prader-Willi Syndrome
  2.4.2. Angelman Syndrome
  2.4.3. Beckman Weidworth Syndrome

UNIT-III

Human Heredity and Social Welfare

3.1. Color, Form and Distribution of hair on the head.
3.2. Color and Vision of Eye and Vision defects.
3.3. Shape and size of hands & Feet and their abnormalities.
3.4. Abnormalities and inheritance patterns of Skin, Muscles & Bones.
3.5. Eugenics
  3.5.1. Definition
  3.5.2. History
  3.5.3. Positive and Negative Eugenics
3.6. Euphenics.
3.7. Euthenics.
3.8. Heredity of Twins.

UNIT-IV

Autoimmunity

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 (covered)

Applied Cytogentics

5.1. FISH and its clinical application
5.2. Cytogenetics in Medicine
5.3. Chromosome Mapping in Humans
5.4. Analysis of Mitotic Chromosomes

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

Major test:

i. Major test will have seven questions each of 15 marks
ii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus
iii. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

Books Recommended:

1. Chromosome Abnormalities and Genetic Counselling (Oxford Monographs on Medical Genetics) R.J. McKinlay Gardner, Grant R.Sutherland, 3 edition, 2003. Oxford University Press, USA.
2. Human Chromosomes by Orlando J.Miller and Eeva Therman, 2000, Publisher Springer Us.
3. De Grouchy & Turleau.1984. Clinical atlas on Human Chromosomes.
4. Human Heredigy : Principles and Issues by Micheal R. Cummings; 4th edition, 1997, West Group.
5. Jankowski & Polak, 1996.Clinical Gene Analysis and Manipulation.
6. Rimoin *et al, 2002.Principles* & Practice of Medical Genetics, vol. I-III
7. Robinson and Linden,1994. Clinical Genetics Handbook.
8. F Vogel A.G. Motulusky. Human Genetics: Problems and Approaches. Second Completely Revised Edition, Springer-Verlag.
9. Golder N. Wilson, M.D., Ph.D.Clinical Genetics-A Short Course. A John Wiley and Sons, Inc., Publication.
10. Human Cytogenetics; General Cytogenetics V.I by John Laurence Hamerton, 1971

Course no. PSHGTC301 **Title: Applied medical genetics**

Credit: 04

Duration of examination: 2 Hrs & 30 mins **Maximum Marks:**

A) Semester examination:60

B) Sessional examination: 40

Minor 1=20

Minor-2=20 Total=40

Applied Medical Genetics

Objective: The course has been designed with the objectives to make the students of human genetics to learn about the importance of Genetics in medicine. An understanding about the applications of principles of Genetics is finally useful for the welfare of family as well as society because the genetic diseases are largely incurable and efforts are made to prevent their transmission.

Unit-I

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

1.4. Evolving molecular cytogenetic technologies

1.4.1. Comparative genomic hybridization

1.4.2. Spectral hybridization

1.5. Human mitochondrial DNA and related diseases

Unit-II

2.1. Inheritance pattern of genetic diseases

2.1.1. Autosomal disorders

2.1.2. X-linked dominant disorders

2.1.2.1. Familial rickets

2.1.2.2. Hereditary nephritis

2.1.3. X- linked recessive disorders

2.1.3.1. Color blindness

2.1.3.2. 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

Unit-III

3.1. Chromosomal abnormalities in human cancers

3.1.1. Chromosomal changes associated with leukemias

3.1.2. Chromosomal changes associated with solid tumors

3.1.3. Chromosomal associated with benign tumors

3.2 Autosomal dominant disorders with the predisposition to develop cancer

3.3. Association of HPV with human cervical carcinoma

Unit-IV

4.1. Prenatal testing and reproductive genetics

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. Mechanism of Gene silencing

Unit-V

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
    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

Major test:

i. Major test will have seven questions each of 15 marks
ii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus
iii. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

Books Recommended:

1. Cox & Sinclair, Molecular Biology in Medicine, Blackwell, Latest Edition.
2. DeGrouchy & Turleau, Clinical Atlas on Human Chromosomes, Wiley, Latest Edition.
3. Jankowski & Polak, Clinical Gene Analysis and Manipulation , Cambridge, Latest Edition.
4. Korf, Human Genetics- A Problem Based Approach, Blackwell, Latest Edition

Course No. PSHGTC- 302
Credits: 04
Duration of examination: 2 Hrs & 30 mins

Title: Medical Biotechnology

Maximum Marks:

A) Semester examination:60
B) Sessional examination: 40
Minor 1=20
Minor-2=20 Total=40

Medical Biotechnology

Objective: Advances made in the area of biotechnology are being used in the field of medicine for the diagnosis of the human genetic diseases. 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. Besides this, the role of Gene therapy and Stem cell therapy has also been included in the course.

Unit-I

Diagnostics

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, X-ray, TIFA, maternal fetal serum and fetal cells in the maternal blood.

Unit-II

Therapeutics

2.1. Gene therapy

2.1.1. Ex-vivo, In vivo, In situ gene therapy

2.1.2. Stratagies of gene therapy: ADA deficiency, CFTR

2.1.3. TFO, antisense therapy, Ribozymes, Protein aptamers, Intrabodies

2.2. Vectors used in gene therapy

2.2.1. Biological vectors- retrovirus, adenovirus, herpes

2.2.2. Synthetic vectors- liposomes, receptor mediated gene transfer

2.3. Gene therapy trials- Familial hypercholesterolemia, cystic fibrosis and solid tumors.

Unit-III

Therapeutics

3.1. Stem cells and stem cell therapy

3.1.1. Embryonic and adult stem cells

3.1.2. Characteristics of stem cell

- Totipotent cells
- Pluripotent cells
- Multipotent cells

3.2. Embryonic stem cell culture

3.3. Adult stem cell culture and differentiation

3.4. Human cord blood stem cell isolation

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

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- herpes simplex virus, hepatitis

4.2.2. Attenuated vaccines

4.3. Peptide based drugs

Unit-V

Medicine and the future

5.1. Pharmacogenetics

5.2. pharmacogenomics

5.3. Nanomedicine

5.3.1. Nanoparticles

5.3.2. Nanodevices- Medical microrobotics and Nanorobotics

5.3.3. Nanomedicine and nanosurgery- for cancers, neurological disorders

5.4. Molecular modeling

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

Major test:

i. Major test will have seven questions each of 15 marks

ii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus

iii. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

Books Recommended:

1. Cox & Sinclair, Molecular Biology in Medicine, Blackwell, Latest Edition.
2. Pasternak, An Introduction to Molecular Human Genetics, Fritzgarald, 2000.
3. Strachan & Read, Human Molecular Genetics, Wiley, Latest Edition.
4. Rasko & Downes, Genes in Medicine, Klumer, Latest edition.
5. Rimion et al., Principles and Practice of Medical Genetics, Vol-I-III, Churchill, 2002.
6. Robinson and Linden, Clinical Genetics handbook, Blackwell, latest edition.
7. Judit Pongracz, Mary Keen, Medical Biotechnology, 2009.
8. Albert Sasson, Medical Biotechnology: Achievements, Prospectus and Perceptions, 2006.

Course no. PSHGTC- 303

Title: Population genetics, Developmental Genetics and Human Parasitology

Credit: 04

Duration of examination: 2 Hrs.& 30 mins

Maximum Marks:

a) Semester examination: 60

b) Sessional examination: 40

Minor 1=20

Minor-2=20 Total=40

Objective: The course is designed with the objective to provide sound knowledge to the students about the principles of population genetics and evolution and to understand the role of genetics in human reproduction and development. The course will deal with the association of parasites with the human infections. Therefore, the students of human genetics will also learn about the parasites, their life cycle and pathogenicity.

Unit-I

1.1. Drawing and interpreting pedigrees

1.2. Hardy Weinberg Law

1.3. Genetic Polymorphism

1.4. Phenotypic plasticity

Unit-II

2.1. Inbreeding, Inbreeding depression and Heterosis
2.2. Genetic drift
2.3. Genetic variation: its origin and detection
    2.3.1. Mutation- the source of genetic variations
    2.3.2. Detection and measurement of genetic variations
2.4. Behavioral Genetics
    2.4.1. Schizophrenia
    2.4.2. Bipolar disorders
    2.4.3. Alcoholism

Unit-III

3.1. Developmental gene families
3.2. Limb as a development model
3.3. Development gene and cancer
3.4. Sexual differentiation and determination
3.5. Hydatiform moles

Unit-IV

4.1. Male and Female reproductive systems
    4.1.1. Gonads and differentiation of reproductive systems
    4.1.2. Hormonal regulation of sexual differentiation
4.2. Reproductive disorders
    4.2.1. Pseudohermaphroditism
    4.2.2. True hermaphroditism
    4.2.3 Gonadal dysgenesis
    4.2.4. Anomalies of genital ducts
    4.2.5. Recurrent pregnancy loss
4.3. Assisted reproductive technologies

Unit-V

5.1. Scope and definition of parasitilogy
5.2. Protozoan parasites
    5.2.1. Plasmodium
    5.2.3. Entamoeba
5.3. Helminth parasites
    5.3.1. Taenia *solium*
    5.3.2. Wuchereria *bancrofti*
5.4. GIT as a habitat of protozoan and helminth parasities of man
5.5. Blood and lymph as habitat of parasite
5.6. Reticuloendothelial system as habitat of parasite of a man

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

Major test:

i. Major test will have seven questions each of 15 marks

ii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus

iii. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

Books Recommended:

1. F Vogel A.G. Motulusky. Human genetics: Problems and Approaches. Second completely revised edition, Springer- Verlag.
2. Golder N, Wilson M.D., Ph.D. Clinical Genetics- A short course, A John Wiley and Sons, Inc., Publication.
3. Elaine Johanson Mange and Arthur P. Mange. Basic Human Genetics. Rastogi Publications.
4. Ricki Lewis. Human Genetics- Concepts and Application, second edition. WCB- McGraw Hill.
5. Parasitology by K.D. Chatterjee, Calcutta: Chatterjee Medical Publishers, Latest edition.
6. Medical Zoology: An Introduction to Parasitology by R.C. Sobti, Published by M/s Shobhan Lal and company, Latest Edition.
7. Emery's Medical Genetics, Latest Edition
8. Principles of Genetics by Simmons and Snustad, Hoboken: Wiley, 2009, 5^{th} edition.

Course No. PSHGTE304 **Course Title: Human Genetic Disorders and Society**

Credits: 4

Unit-I- Chromosomes and Cell division

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

Unit-II - Chromosome Disorders

2.1 Incidence of chromosome abnormalities

2.2. Disorders of the Autosomes

2.2.1. Down syndrome

2.2.2 Edward syndrome

2.2.3. Patau syndrome

2.3. Disorders of the sex chromosomes

2.3.1. Turner syndrome

2.3.2. Klinefelter syndrome

2.4. Sex limited, sex linked and sex influenced traits

Unit-III- Human Genome

3.1. Central Dogma of Life

3.2. DNA- the Genetic Material of Human

3.3. DNA Diagnostic Techniques

3.3.1. PCR

3.3.2. DNA fingerprinting

3.4. Genetic testing

3.4.1. Pre natal DNA diagnosis

3.4.2. Pre-implantation genetic testing

3.4.2. Carrier detection

Unit-IV- 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- Genetics of Multifactorial diseases

5.1. Diabetes mellitus

5.2. Cardiovascular diseases

5.3. Hyperthyroidism

5.4. Schizophrenia

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

Major test:

i. Major test will have seven questions each of 15 marks

ii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus

iii. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

Books recommended:

8. Alberts et al, 2002.Molecular Biology of the Cell
9. De-Robertis, 2003. Cell & Molecular Biology (latest Edition)
10. Ricki Lewis. Human Genetics- Concepts and Application, second edition. WCB- McGraw Hill.
11. DeGrouchy & Turleau, Clinical Atlas on Human Chromosomes, Wiley, Latest Edition.
12. Strachan & Read, Human Molecular Genetics, Wiley, Latest Edition.
13. Rimion et al., Principles and Practice of Medical Genetics, Vol-I-III, Churchill, 2002
14. Emery's Elements of Medical Genetics, 14th edition by Drs. Peter Turnpenny and Sian Ellard.

Course No. PSHGTC-401 **Title: - Genetic Counselling**
Credits: 04
Duration of Examination: 2 Hrs & 30 mins **Maximum Marks:**
a) Semester Examination: 60
b) Sessional Assessment: 40
Minor 1=20
Minor-2=20 Total=40

Genetic Counselling

Objectives: - 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. It will also help in providing the necessary information on support groups and services to manage the genetic condition.

Unit-I

Genetic aspects of Genetic Counselling

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
1.6 Risk communication a complex process

Unit-II

Genetic counseling in specific organ system genetic disorder

2.1 Neuromuscular diseases
2.2 Central nervous system disorders
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

Genetics and Society

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

Unit-IV

4.1. Abortion as a method fertility control.
4.2 Status of an unborn child
4.3 Female foeticide in India
4.4 Relationship between PCP&DT Act 1994 and MPT Act 1971

Unit-V

5.1. Genetic counseling strategies for working with families

5.2. Developmentally based approaches for counseling children and adolescents.
5.3. Genetic counseling for women with intellectuals disabilities.
5.4. Actively engaging with patients in decision making.

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

Major test:

vii. Major test will have seven questions each of 15 marks

viii. One question will be very short answer type of multiple parts compulsory spread over entire syllabus

ix. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

Books Recommended:

1. Baker et al., A guide to Genetic Counseling, Wiley, 1998.
2. Rose and Lucassen, Practical Genetics of primary case, Oxford, 1999.
3. Young, Introduction to Risk Calculation in Genetic Counselling, Oxford, 1999.
4. Genetics in practice: A clinical approach for Healthcare Practitioners by Jo Hayden.
5. Lessons learned: Risk Management issues in Genetic Counseling by Susan Schmerler.
6. Introduction to Risk Calculation in Genetic Counseling by Ian Young.
7. Genetic Couselling: a psychological approach by Christine Evans

Chromosome abnormalities and genetic counseling by Grant R Sutherland, R. J. M. Gardner.

Course No. PSHGTC-402
Credits: 04
Duration of Examination: 2 Hrs & 30 mins

Title: - Clinical Genetics

Maximum Marks:
a) Semester Examination: 60
b) Sessional Assessment: 40
Minor 1=20
Minor-2=20 Total=40

Clinical Genetics

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

UNIT-I

1.1 Principles and practice of clinical genetics
1.2 History , Nature and frequency of genetic diseases
1.3 Molecular and Biochemical basis of genetic diseases
1.4 Late onset genetic disorders- Alzheimer
1.5 Multifactorial diseases- Atherosclerosis, Diabetes mellitus
1.6 Nature and Nurture: Distangling the effects of genes and environment

UNIT-II

2.1 Cytogenetic techniques in disease detection
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
- **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

Case studies: Autosomal abnormalities

4.1. Introduction to Autosomal abnormalities
4.2. Down syndrome
4.3. Patau syndrome
4.4. Edward syndrome
4.5. Incidence of Autosomal disorders
4.6. Double Trisomy

UNIT – V

Case Studies: Sex Chromosomal Abnormalities

5.1. Introduction to sex chromosomal abnormalities

5.2. Klinefelter Syndrome
5.3. Turner syndrome
5.4. Primary Amenorrhea
5.5. Ambiguous genitalia
5.6. Hypospadias
5.7. Intersex

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

Major test:

Books Recommended:

1. Emerys Medical Genetics by Robert F. Mueller, Ian D. Young, Publisher Churchill Livingstone, 13th Edition.
2. Principles of Genetics by D. Peter Snustad & Michael J siimons, fourth edition.
3. ABC of clinical genetics by Helen m Kingston, publisher Springer Berlin/ Heidelberg, third edition.
4. Clinical genetics: source book for physicians (a Wiley medical publication) by Laird G. Jackson and R. Neil Schimke (hardcover- Oct 1979

Course No. PSHGTO-403 **Title: Biotechnology & Health Care**
Credits: 04
Duration of Examination: 2 Hrs & 30 mins **Maximum Marks:**

a) Semester Examination: 60 b) Sessional Assessment: 40
Minor 1=20
Minor-2=20 Total=40

Unit-I

1.1. Origin and definition of biotechnology

1.2. Scope and importance of biotechnology

1.3. Recombinant DNA

1.4. Gene cloning

1.5. Cloning vectors

Unit-II

2.1. Animal cell and tissue culture

2.2. Advantages and disadvantages of tissue culture

2.3 Culture media for cell and tissue

2.4. Human lymphocyte culture

2.5. Bone marrow culture

Unit-III

3.1. PCR and its principle

3.2. Basic PCR and its modification: Inversed PCR and anchored PCR

3.3. Applications of PCR in biotechnology

3.4. DNA fingerprinting

3.5. Uses of DNA fingerprinting

Unit-IV

4.1. Immune System and its components

4.2. Immune Response

4.3. Antibodies and Antigens

4.4. Production of vaccines

4.5. In vitro fertilization in humans

Unit V

5.1. Environmental components

5.2 Environmental pollution and its types:

5.2.1 Water pollution

5.2.1. Soil pollution

5.3. Non renewalble resources and renewable sources of energy

5.4. Biofertilizers

5.5. Biopesticides

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

Major test:

iv. Major test will have seven questions each of 15 marks

v. One question will be very short answer type of multiple parts compulsory spread over entire syllabus

vi. The remaining six questions will be from the remaining 41% to 100% portion of the syllabus and the candidate will have to attempt any three of them

Books Recommended:

9. Cox & Sinclair, Molecular Biology in Medicine, Blackwell, Latest Edition.
10. Pasternak, An Introduction to Molecular Human Genetics, Fritzgarald, 2000.
11. Strachan & Read, Human Molecular Genetics, Wiley, Latest Edition.
12. Rasko & Downes, Genes in Medicine, Klumer, Latest edition.
13. Rimion et al., Principles and Practice of Medical Genetics, Vol-I-III, Churchill, 2002.
14. Robinson and Linden, Clinical Genetics handbook, Blackwell, latest edition.
15. Judit Pongracz, Mary Keen, Medical Biotechnology, 2009.
16. Albert Sasson, Medical Biotechnology: Achievements, Prospectus and Perceptions, 2006.