

# SUMANDEEP VIDYAPEETH

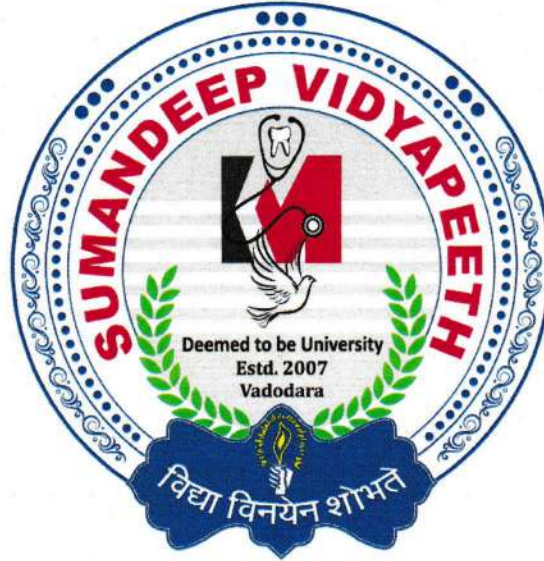
(Declared as Deemed to be University under Section 3 of the UGC Act 1956)

Accredited by NAAC with a CGPA of 3.53 out of four-point scale at 'A' Grade

At & Post Piparia, Tal: Waghodia 391760 (Gujarat) India.

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## CURRICULUM

### Master of Science (M.Sc) HUMAN GENETICS

Attested CTC

*Sharaney*  
15/2/2021

Vice-Chancellor

Sumandeep Vidyapeeth

An Institution Deemed to be University

Vill. Piparia, Taluka: Waghodia.

Dist. Vadodara-391 760. (Gujarat)

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### 1.) Goal:

The main goal of the post graduate education in Human Genetics is to enable a student understand, envisage and explain genetic processes as molecular events and apply his knowledge and skills in clinical genetic problem solving and scientific research.

### 2.) Program Outcomes:

PO-1 To understand and explain genetic processes, various aspects and concepts of Human genome and related genetic disorders with its clinical significance.

PO-2 To acquire skills in techniques used for the genetic testing purposes in order to make genetic testing relevant in day to day life.

PO-3 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.

PO-4 To identify the high risk and low risk individuals for a particular genetic trait and to make concrete proposals with definite outcomes as per defined objectives considering practical aspects of Human Genetics.

PO-5 To design unique research proposal and carry out research study based on Novel Human Genetics principles.

PO-6 To promote health by understanding the genetic basis of common diseases and early detection of potentially treatable genetic conditions, and To spread awareness in the society about these problems for improving human health in rural population.

PO -7 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 The Pre-Conception and Pre-Natal Diagnostic Techniques (Prohibition Of Sex Selection) (PCPNDT) Act while performing genetic counseling and diagnosis.

### 3.) Course Outcomes:

#### Semester-I

1. To understand and describe the structure and function of cell membrane and related transport mechanisms, structure, function of different cell organelles, bio molecules, processes of cell division, mechanism of cellular processes- cell cycle and regulation, Cellular energetic, Signal transduction and Programmed cell death.
2. To understand and describe the basic concepts of Human Skeletal system, the physiology of digestive system, respiratory system, cardiovascular system and nervous system, concepts of Human Endocrinology, concepts of human fertilization and embryonic development.
3. To understand and describe the structure and functions of Nucleic Acid, concepts of Translation and gene regulation, concepts of Human Genome and its evolution, various hybridization assays.
4. To understand and describe basics of cytogenetics and extensions of Mendelism, Structure of Human Chromosomes and related conditions, chromosomal theory of inheritance.



- To understand and describe basic research methodologies - Centrifugation, Electrophoresis and microscopy, different types of blotting techniques, various types of Chromosomal and Molecular analysis techniques.
- To acquire practical skills related to point no.1-5.

### **Semester-II**

- To understand and describe various Biochemical Metabolic pathways, their genetic considerations and related disorders.
- To understand and describe basics of Microbiology, the structure, shape, nutrition & genetics, pathogenic nature of microbes, immunology in health and diseases.
- To understand and describe principle & process of DNA cloning, pathology of different genetic diseases at molecular level, DNA sequencing, Transcirtptome& protein profiling, different DNA diagnostic approaches.
- To understand and describe Human congenital defects and anomalies, genetic basis of cancer, role of eugenics in society, genetic basis of autoimmune diseases and immnodeficiencies, and basics of advanced cytogenetic techniques.
- To acquire practical skills related to point no.1-4.

### **Semester-III**

- To understand and describe various human mitochondrial diseases, study of human genetic diseases using animal model, inheritance patterns of different genetic diseases, environmental, chromosomal and molecular etiology of Cancer, management of genetic diseases, genetic evaluation and treatment of human infertility.
- To understand and describe research methodologies being applied for the diagnosis of the human genetic diseases, gene therapy and its role in therapeutics, role of Stem cells therapeutics, application of medical biotechnology in synthesis of different vaccines and peptide based drugs, pharmacogenomics and nanotechnology.
- To understand and describe theories of evolution, population genetics, use of bioinformatics in Human Genetics, different biostatistical tools used in analysis of genetic and non-genetic data.
- To understand and describe basics of chromosomes and the various techniques used for chromosome analysis, different disorders of autosomes and sex chromosomes, mechanism of DNA replication and its expression, genetic aspects of different types of cancer,
- To acquire practical skills related to point no.1-4

### **Semester-IV**

- To understand and describe importance of Genetic Counseling in the welfare of family as well as society, different testing issues viz. discrimination, privacy and confidentiality, issues associated with human cloning, organ transplantation & Surrogacy, Pre-conception and Pre-natal Diagnosis Act in Human Genetics.
- To understand and describe role of clinical genetics, rerogenetics, treatment of genetic diseases, etiology and genetics of multifactorial diseases, rare genetic diseases.
- To understand and describe recent advances in the field of molecular genetics & biotechnology, single gene disorders, different techniques used in detecting genetic abnormalities,
- To acquire practical skills related to point no. 1-3.
- To carry out research project on Human Genetics.



#### 4.) Admission:

##### 4.1 Eligibility

- The student is required to obtain at least 50% in his/her Bachelor's Programme.
- Bachelor's degree in any branch of science/medicine (with degrees such as B.Sc., B.Pharm, MBBS, BDS, BPT, BAMS, BHMS) from an Institute recognized by respective Statutory Council/ UGC.
- The eligible subject areas are: Life sciences (zoology, botany, genetics, human biology, general life sciences, ecology, environmental biology), bioinformatics, biotechnology, chemistry, pharmaceutical sciences, medicine, dentistry, AYUSH.
- There is no age bar for applying.

##### 4.2 Fee structure and Reservation

- As laid down by Sumandeep Vidyapeeth Deemed to be University.

#### 5.) Course Content

##### 5.1 Semester-I

	Theory	Teaching Hours/ week
Subject Code	Subject	
MHG101	Cell Biology	4
MHG102	Human Physiology	4
MHG103	Human Molecular Genetics-I	4
MHG104	Principles of Human Genetics	4
MHG105	Genetic diagnostics & Human Health Care	4
	<b>Practical</b>	
MHG106	Practicals based on Theory MHG101-102	4
MHG107	Practicals based on Theory MHG103-105	4
	<b>Total Hours</b>	<b>28</b>

##### 5.2 Semester-II

	Theory	Teaching Hours/ week
Subject Code	Subject	
MHG201	Biochemistry of Metabolic Disorders	4
MHG202	Clinical Microbiology & Immunology	4
MHG203	Human Molecular Genetics-II	4
MHG204	Human Cytogenetics	4
	<b>Practical</b>	
MHG205	Practicals based on Theory MHG201-202	6
MHG206	Practicals based on Theory MHG203-204	6
	<b>Total Hours</b>	<b>28</b>

##### 5.3 Semester-III

	Theory	Teaching Hours/ week
Subject Code	Subject	
MHG301	Applied Medical Genetics	4



MHG302	Medical Biotechnology	4
MHG303	Population genetics, Bioinformatics & Biostatistics	4
MHG304	Human Genetic Disorders & Society-I	4
	<b>Practical</b>	
MHG305	Practicals based on Theory MHG301-302	6
MHG306	Practicals based on Theory MHG303	4
	<b>Total Hours</b>	<b>26</b>

#### 5.4 Semester-IV

	Theory	Teaching Hours/ week
Subject Code	Subject	
MHG401	Clinical Genetics	4
MHG402	Human Genetic Disorders & Society-II	2
MHG403	Research Project and Dissertation work	12
MHG404	Genetic Counseling	4
	<b>Practical</b>	
MHG405	Practicals based on Theory MHG401-402	6
	<b>Total Hours</b>	<b>28</b>

## 6.) Syllabus

### 6.1 Semester-I

MHG101: Cell Biology

#### Chapter-1 Cell: The basic unit of life

1.1 Cell: Structure and Organization

1.2 Plasma Membrane

1.2.1 Structure of Plasma Membrane with special emphasis on various models

1.2.2 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

#### Chapter-2 Understanding Cell Organelles

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

3 Structure and Function:



- 2.3.1. Nucleus
- 2.3.2. Ribosomes
- 2.3.3. Lysosomes
- 2.3.4. Peroxisomes

### **Chapter-3 Cell division**

- 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

### **Chapter-4 Cellular interactions and Cell Cycle**

- 4.1 Cell-Cell Interaction
  - 4.1.1 Cell adhesion molecules
  - 4.1.2 Cellular Junctions
  - 4.1.3 Extracellular matrix
- 4.2 Cell cycle and its regulation
  - 4.2.1 Cyclin and Cyclin dependent kinases
  - 4.2.2 Centrosome Cycle
  - 4.2.3 Cell cycle checkpoints
  - 4.2.4 Role of Rb and p53 protein in cell cycle regulation

### **Chapter-5 Cell Signaling**

- 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, Phosphatidylinositol 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 signaling
- 5.7 Programmed cell death (Apoptosis)

### **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. Cooper 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, 6<sup>th</sup> Edition, Taylor & Francis Group, 2014.
- 7) Gerald Karp. Cell and Molecular Biology: Concepts and Experiments, 8<sup>th</sup> Edition John Wiley and Sons, 2016.
- 8) Rastogi, V.B. Cell Biology. Third Edition. New Age International Publishers, 2016



MHG102: Human Physiology

**Chapter-1 Skeletal system**

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

**Chapter-2 Human Systems and their Physiology– I**

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

**Chapter-3 Human Systems and their Physiology– II**

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

**Chapter-4 Human Systems and their Physiology– III**

4.1 Gross anatomy of Human Excretory system

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

4.2 Physiology of Endocrine System

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

**Chapter-5 Human Embryology**

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

**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, 13<sup>th</sup> edition, 2015.
- 4) Inderbir Singh, Human Embryology, 11<sup>th</sup> edition, 2017,
- 5) GK Pal, Medical Physiology, 13<sup>th</sup> Edition, Orient Black Swan, 2018.
- 6) K Sembulingam, Essentials of Medical Physiology, 8<sup>th</sup> Edition, JAPI Brothers Medical Publishers, 2019.
- 7) Guyton and Hall. Text book of Medical Physiology. 12<sup>th</sup> Edition. Elsevier Saunders Publishers, 2019.

MHG103: Human Molecular Genetics

**Chapter-1 Human Genome**

- 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

**Chapter-2 Nucleic Acid: Structure & functions**

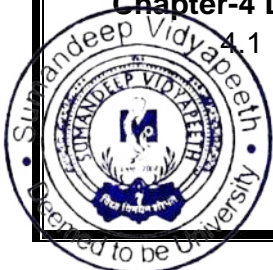
- 2.1 DNA – Structure & types
- 2.2 DNA Replication in Prokaryotes & Eukaryotes.
- 2.3 RNA – Structure & types
- 2.4 Mechanism of Transcription and transcription factors
- 2.5 Post-transcriptional modification:
  - 2.5.1 5' Capping
  - 2.5.2 Polyadenylation
  - 2.5.3 Splicing
  - 2.5.4 RNA editing

**Chapter-3 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

**Chapter-4 DNA Mutations**

- 4.1 Mutations
  - 4.1.1 Types of mutations





#### 4.1.2 Physical & Chemical Mutagens

4.2 Amestest

4.3 Complementation test

4.4 DNA Repair

4.4.1 DirectRepair

4.4.2 Excision Repair (Base Excision Repair, Nucleotide Excision Repair, Mismatch Repair, Non homologous end joining, SOS repair)

### **Chapter-5 Molecular Hybridization**

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

### **Books Recommended:**

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

MHG104: Principles of Human Genetics

### **Chapter-1 Hereditary and Variations**

1.1 History of Cytogenetics

1.2 Mendel's Laws of Heredity: Law of segregation & Law of Independent Assortment

1.3 Test Cross & BackCross

1.4 Extensions of Mendelism:

1.1.1 Incomplete Dominance

1.1.2 Co-dominance

1.1.3 Multiple allelism

1.1.4 Epistasis

1.1.5 Pleiotropy

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

### **Chapter-2 Introduction to Chromosomes**

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 & Klinefelters syndrome

## Chapter-3 Chromosomal & Non-chromosomal basis of sex determination

- 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

### Books Recommended::

- 1) F Vogel A.G. Motulsky. Human Genetics: Problems and Approaches. 5<sup>th</sup> Edition, BMC, 2010.
- 2) Ricky Lewis, Concepts of Human Genetics, 2011.
- 3) Bruce R. Korf and Mira B Irons, Human Genetics & Genomics (4<sup>th</sup> edition), 2013.
- 4) ABC of Clinical genetics, Helen M Kingston, 4<sup>th</sup> Edition, BMJ, 2015.
- 5) Robert Nussbaum et al. Thompson & Thompson genetics in Medicine, 8<sup>th</sup> Edition, Elsevier, 2015.
- 6) Robert L. Nussbaum, Roderick R. McInnes, & Huntington F. Willard, Thompson & Thompson Genetics in Medicine, 8<sup>th</sup> edition, Imprint : Saunders, 2015.
- 7) Human Heredity: Principles and Issues by Micheal R. Cummings; 11<sup>th</sup> edition, Cengage Learning, 2016.
- 8) Emerys & Rimoim, Principles & Practice of Medical Genetics, 7<sup>th</sup> Edition, Elsevier, 2017.

MHG105: Genetic diagnostics & Human Health Care

### Chapter-1 Analytical Techniques

- 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.

### Chapter-2 Blotting & Microscopy

- 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.

### **Chapter-3 Molecular Diagnostic Techniques**

- 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

### **Books Recommended:**

- 1) David LS peeta and Bobert D,Basic Method in Microscopy,2006.
- 2) Cox & Sinclair, Molecular Biology in Medicine,Blackwell,..2009.
- 3) De Grouchy & 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, 11<sup>th</sup> edition.WCB-McGrawHill,2011.
- 7) Andreas Hofmann and Samuel Clokie, Wilson and Walker's Principles and Techniques of Biochemistry and Molecular Biology, Cambridge University Press Edition 8<sup>th</sup>,2018.

MHG106: Practicals based on Theory MHG101-102

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 Transverse Section of Human Pancreas, Thyroid follicles, Ovary, Sperm, Adrenal gland and Testis.
7. Identify and study the different types of Bones- Long & Short bones.
8. To study the different types of girdles-Pectoral &Pelvic.
9. To study the different bones of Human Skull.
10. To study the different types of joints in Humans.



11. To measure blood pressure by Sphygmomanometer.
12. To measure Mean arterial pressure and pulse pressure estimation.

MHG107: Practical based on Theory MHG103-105

1. To study the different biosafety levels and good lab practices.
2. To perform Sterilization of glassware and plastic ware.
3. To study the study the presence of Drumstick in human neutrophil 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
  - 8.1. Down syndrome
  - 8.2. Edward syndrome
  - 8.3. Patau syndrome
  - 8.4. Turner syndrome
  - 8.5. 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.

## 6.2 Semester-II

MHG201: Biochemistry of Metabolic Disorders & Developmental genetics

Chapter-1 Carbohydrates Metabolism

1. Introduction to Carbohydrates Metabolism
- Disorders of Carbohydrates Metabolism (Genetic cause, diagnosis & treatment):



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

### **Chapter–2 Protein Metabolism**

- 2.1 Introduction to Proteins & Amino acids.
- 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 Erythropoietin Porphyria
- 2.5 Metabolic disorders of Glycosaminoglycans & Glycoproteins (Genetic cause, diagnosis & treatment):
  - 2.5.1 Mucopolysaccharidosis
  - 2.5.2 Mucopolidosis

### **Chapter–3 Lipid Metabolism**

- 3.1 Introduction to Lipids & Fatty acids &their metabolism
- 3.2 Disorders of Lipid Storage (Genetic cause, diagnosis &treatment):
  - 3.2.1 Tay Sachs Disease
  - 3.2.2 Krabbe Disease
- 3.3 Disorders of Fatty acid Metabolism (Genetic cause, diagnosis &treatment):
  - 3.3.1 Hyperlipidemia
  - 3.3.2 Hypercholesterolemia

### **Chapter-4 Developmental Genetics**

- 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

### **Chapter-5 Reproduction Biology**

- 5.1. Male and Female reproductive systems
  - 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. Truehermaphroditism
  - 5.2.3. Gonadal dysgenesis
  - 5.2.4. Anomalies of genital ducts



### **Books Recommended:**

- 1) T. Subramaniam Molecular Developmental biology, 2008.
- 2) Mathews et al.: Biochemistry (4rd Ed.), Pearson, 2012.
- 3) Harpers illustrated Biochemistry (31st edition) The McGraw Hill Companies, 2014.
- 4) Berg et al.: Biochemistry (8th Ed.), Freeman, 2015.
- 5) Biochemistry by Donald Voet (5th edition) publisher: 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 McGraw Hill Companies, 2018.
- 9) Harpers illustrated Biochemistry (31st edition) The McGraw Hill Companies, 2018.

MHG202: Clinical Microbiology & Immunology

### **Chapter-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: their shapes and type
- 1.3 Microbial nutrition (Auxotroph's, 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

### **Chapter –2 Microbial diseases - Etiology, Pathogenesis and their Control**

- 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 Diarrhea, Salmonella infection and food poisoning.
- 2.3 Viral diseases:
  - 2.3.1 AIDS
  - 2.3.2 Hepatitis
  - 2.3.3 H1N1 Infection

### **Chapter-3 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 antigenicity
- 3.3 Structure of T & B cells
- 3.4 Immunoglobulin's: Types, Structure & Function
- 3.5. Cytokines, their role in human diseases

### **Chapter-4 Immunology-II**

- 4.1 Major Histocompatibility complex
- 4.2 Role of HLA in disease susceptibility
- 4.3 Transplantation immunology (Allograft, Xeno graft, Syngraft, graft, graft rejection)  
Antigen processing & presentation



- 4.5 T-cell maturation, activation and differentiation
- 4.6 B-cell maturation, activation and differentiation

### **Chapter-5 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

### **Books Recommended:**

- 1) Richard Coico, Geoffrey Sunshine, Immunology (A short course), 6<sup>th</sup> 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. Goldsby, 7th edition, 2013.
- 4) Prescott, Harley, Klein; Microbiology, 10th edition, McGraw- Hill Higher Education, 2017.
- 5) Pelczar, Michael J. Jr. / Chan, E.C.S / Krieg, Noel R., Microbiology, 5th Edition, McGraw-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., Janeway's Immunobiology, 9th GS publications, 2019.

MHG203: Human Molecular Genetics-II

### **Chapter-1 DNA Cloning**

- 1.1. Cell -based DNA cloning
  - 1.1.1. Principles of DNA cloning
  - 1.1.2. Cloning vectors: Plasmids, Phages, Cosmids, YAC
- 1.2. Expression cloning
- 1.3 Cell free DNA cloning
  - 1.3.1. Principles of PCR
  - 1.3.2. PCR types and its applications
- 1.4. DNA Fingerprinting

### **Chapter-2 Molecular Pathology**

- 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.
- Molecular pathology of chromosomal disorders.
- Epigenetics and its role in Human diseases.



## **Chapter – 3 Genetic Testing in Individuals and Populations**

- 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 formulation.
- 3.5. Testing for a specified sequence change

## **Chapter-4 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 (2D Electrophoresis & MALDI-TOF)
  - 4.4.2 Identifying Proteins that interact with one another (Yeast hybrid system, phage display)
  - 4.4.3 Protein degradation.

## **Chapter-5 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-Syndrome
  - 5.2.6 Cystic Fibrosis

### **Books Recommended:**

- 1) F Vogel A.G. Motulsky. Human Genetics: Problems and Approaches. 4<sup>th</sup> 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, Genomes 4, 4th edition, Garland Science, 2017.
- 8) Lewin, Gene XII, 12th Edition, 2017.

MHG204: Human Cytogenetics

## **Chapter – 1 Human Congenital Anomalies**

- 1.1 Introduction
- 1.2 Neural Tube Defects
- 2.1 Anencephaly





- 1.2.2 Encephalocele
- 1.2.3 Hydrocephaly
- 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

## **Chapter-2 Cancers**

- 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 (Burkett's Lymphoma & Philadelphia chromosome).
- 2.6 Involvement of TSGs in;
  - 2.6.1. Retinoblastoma
  - 2.6.2. Breast cancer

## **Chapter-3 Chromosome Instability Syndrome and Eugenics**

- 3.1. Chromosome Instability syndrome: Ataxia Telangiectasia, Fanconi Anemia.
- 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. Euthenics, Euthenics and Euthanasia

## **Chapter-4 Autoimmune Diseases**

- 4.1. Genetic Basis of Autoimmune Diseases
  - 4.1.1 Rheumatoid Arthritis
  - 4.1.2 Grave's 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 and Leukemia

## **Chapter-5 Advanced Cytogenetics**

- 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

### **Books Recommended:**

- 1) F Vogel A.G. Motulsky. Human Genetics: Problems and Approaches. 5<sup>th</sup> 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 Counseling (Oxford Monographs on Medical Genetics), 5th edition,. Oxford University Press, USA.2018.

**MHG205: Practicals based on Theory MHG201-202**

1. To study the principle, construction and uses of Laminar airflow.
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:
  - 4.1. Semi-auto analyzer
  - 4.2. Fully auto analyzer
5. Qualitative and quantitative estimation of carbohydrates, Lipids and proteins.
6. Plasma and serum isolation.
7. To perform liver function test of a given serum/blood sample on 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.
5. To study the common apparatus used in microbiology.
6. To study the general morphology of bacteria.
7. To prepare chemicals for bacterial staining.
8. To prepare a temporary mount of bacteria (lactobacillus) present in curd.
9. To prepare the different culture media.
10. To carry out various biochemical tests for Lactobacillus bacteria-
  - 10.1 Gram Staining
  - 10.2 Oxidase Test
  - 10.3 Catalase Test
11. To observe the presence of any bacteria belonging to Enterobacteriaceae family-
  - 11.1 Indole Test
  - 11.2 Methyl Test
  - 11.3 Voges-Proskauer Test
  - 11.4 Citrate Test
  - 11.5 IMViC Test
12. To carry out the extraction of bacterial DNA
13. To carry out the amplification of bacterial DNA by Polymerase Chain Reaction
14. To study the abnormal conditions of genital duct (Hypospadias, Anorectal malformations, Ambiguous genitalia).

**MHG206: Practicals based on Theory MHG203-204**

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:
  - 9.1. Graves disease
  - 9.2. Edward syndrome
  - 9.3. Arthritis
  - 9.4. Cleft lip
  - 9.5. Cleft palate
  - 9.6. Neural tube defects
  - 9.7. Fragile X syndrome
  - 9.8. Hemophilia
10. To prepare karyotype for the provided metaphase plates and identify the genetic condition:
  - 10.1. Down' syndrome
  - 10.2. Turner syndrome
  - 10.3. Klinefelter syndrome
  - 10.4. Patau syndrome
  - 10.5. 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.

### 6.3 Semester-III

MHG301: Applied Medical Genetics

#### Chapter-1 Medical Genetics

- 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

#### Chapter-2 Studying Inheritance Patterns

- 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
    - 2.2.1. Familial rickets
    - 2.2.2. Hereditary nephritis
  - 2.3. X- linked recessive disorders
    - 2.3.1 Colorblindness
    - 2.3.2 Muscular dystrophies- BMD &DMD
- Hemoglobin and Hemoglobinopathy



- 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

### **Chapter-3 Chromosomal abnormalities in human cancers**

- 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

### **Chapter-4 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

### **Chapter-5 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 (nongenetic)
  - 5.2.1 Male
  - 5.2.2. Female
- 5.3 Genetic evaluation of the
  - 5.3.1. Infertile male
  - 5.3.2. Infertile female
  - 5.3.3. Treatment of infertilities

### **Books Recommended:**

- 1) F Vogel A.G. Motulsky. Human Genetics: Problems and Approaches. 5<sup>th</sup> Edition, BMC, 2010.  
Helen M Kingston, ABC of Clinical genetics, , 4<sup>th</sup> Edition, BMJ, 2015.
- 2) Robert Nussbaum et al. Thompson & Thompson genetics in Medicine, 8<sup>th</sup> Edition, Elsevier, 2015
- 3) Micheal R. Cummings Human Heredity: Principles and Issues; 11<sup>th</sup> edition, 2016,
- 4) Emerys & Rimoim, Principles & Practice of Medical Genetics, 7<sup>th</sup> Edition, Elsevier, 2017

MHG302: Medical Biotechnology

### **Chapter-1 Diagnostics**

- 1.1. DNA diagnostics
- 1.2. Biochemical diagnostics
- 1.3. Immunodiagnostics



#### 1.4. Prenataldiagnostics

- 1.4.1 Invasive techniques- Amniocentesis, Fetoscopy, Chorionic VilliSampling
- 1.4.2 Non- invasive techniques- Ultrasonography, maternal fetal serum and fetal cells in the maternal blood.

### **Chapter-2 Therapeutics**

- 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

### **Chapter-3 Stem cells and their applications**

- 3.1. Embryonic and adult stemcells
- 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

### **Chapter-4 Applied Medical Biotechnology**

- 4.1 Gene products inmedicine
  - 4.1.1 Anti- hemophilic factor
  - 4.1.2 Humulin
  - 4.1.3 Erythropoietin
  - 4.1.4 Growth hormone/somatostatin
  - 4.1.5 Interferon
- 4.2 DNA basedvaccines
  - 4.2.1 Subunit vaccines- hepatitis Bvaccine
  - 4.2.2 Attenuatedvaccines
- 4.3 Peptide based drugs

### **Chapter-5 Medicine and the future**

- 5.1. Pharmacokinetics 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

### **Books Recommended:**

1. P. Nallar et al., Medical biotechnology, Oxford Handbooks, 2010.
2. Tom Strachen, Human Molecular Genetics, 4<sup>th</sup> Edition, Garland Science,2010.
3. Surendra Nimesh, Gene therapy: Potential application of Nanotechnology, 1st Edition, Woodhead, 2013.
4. Emerys & Rimoin, Principles & Practice of Medical Genetics, 7<sup>th</sup> Edition, Elsevier,2017.



5. Yui-Wing Francis Lam et al., Pharmacogenomics: Challenges & opportunities in Therapeutic Implementation, 2nd Edition, Academic Press,2018.

**MHG303: Population genetics, Bioinformatics & Biostatistics**

**Chapter-1 Evolution**

- 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

**Chapter-2 Population Genetics**

- 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

**Chapter-3 Behavior Genetics**

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

**Chapter-4 Bioinformatics**

- 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 software's
- 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

**Chapter – 5 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.

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.

**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, 6<sup>th</sup> Edition, Worth Publishers, 2012.
- 4) Arthur M. Lesk, Introduction to Bioinformatics, 4<sup>th</sup> Edition, Oxford, 2013.
- 5) Robert Nussbaum et al. Thompson & Thompson genetics in Medicine, 8<sup>th</sup> Edition, Elsevier, 2015.
- 6) Lisa M. Sullivan, Essentials of Biostatistics in Public Health, 3<sup>rd</sup> Edition, Jones & Bartlett Learning, 2017.
- 7) Jennifer Doudna, Crack in Creation: the new power to control evolution, Vintage Digital, 2017.

MHG304: Human Genetic Disorders & Society-I

**Chapter-1 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
  - 1.4.1 Conventional techniques
  - 1.4.2 Advanced techniques

**Chapter-2 Disorders of autosomes and sex chromosomes**

- 2.1. Disorders of the autosomes
  - 2.1.1. Down syndrome
  - 2.1.2. Edward syndrome
  - 2.1.3. Patau syndrome
- 2.2. Disorders of the sex chromosomes
  - 2.2.1. Turner syndrome
  - 2.2.2. Klinefelter syndrome
- 2.3. Sex limited, sex linked and sex influenced traits

**Chapter-3 Central Dogma of Life**

- 3.1 DNA as a genetic material
- 3.2 DNA Replication
- 3.3 Transcription
- 3.4 Translation

**Chapter-4 Genetics and Cancer**

- 4.1. Genetic and environmental factors in cancers
- 4.2. Genetics of common cancers
  - 4.2.1 Leukemia's
  - 4.2.2 Breast cancer
  - 4.2.3 Cervical cancer
  - 4.2.4 Ovarian cancer



## Chapter-5 Genetics of Multifactorial diseases

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

### 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, 8thEdition, 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, 15thedition , Elsevier, 2017.
- 6) Emerys & Rimoim, Principles & Practice of Medical Genetics, 7th Edition, Elsevier,2017.

### MHG305 Practicals based on Theory MHG301-302

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.

### MHG306: Practicals based on Theory MHG303

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:





- 4.1. Tongue rolling mechanisms
- 4.2. Eye colour
- 4.3. Widow's peak
- 4.4. PTC tasting
5. Study of Hardy Weinberg equilibrium in a given population group.
6. To construct and study phylogenetic trees.
7. To study finger ball 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
  - 9.1. Calculation of central tendencies
  - 9.2. T-test
  - 9.3. Chi square test
  - 9.4. Odd ratio
  - 9.5. ANOVA
  - 9.6. Z-test
  - 9.7. Standard deviation

#### **6.4 Semester-IV**

MHG401: Genetic Counseling

#### **Chapter-1 Genetic Counseling**

- 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

#### **Chapter-2 Genetic Counseling and Genetic Disorders**

- 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

#### **Chapter-3 Issues in Genetic testing-I**

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

#### **Chapter-4 Issues in Genetic testing-I**

- 4.1 Informed Consent and Right of Choice
- 4.2 Human Cloning and Eugenics
- 4.3 Surrogate mothers
- 4.4 Organ banking and transplantation



- 4.5 Medical Ethics in India
- 4.6 Dilemmas faced by Counselors

**Chapter-5 Legal Implications in Genetic Testing**

- 5.1 Pre-natal diagnostic techniques (Regulation and Prevention of Misuse) Act, 1994
  - 5.1.1 Pre conception Pre-natal diagnostic techniques (Prohibition of sex selection) Act
- 5.2 Regulation of prenatal diagnostic techniques
- 5.3 Registration & regulation of genetic counseling centers, genetic laboratories & genetic clinics
- 5.4 Appropriate authority & advisory committee
- 5.5 Offences and Penalties
- 5.6 Medical termination of pregnancy Act

**Books Recommended:**

- 1) Rimion et al., Principles and Practice of Medical Genetics, Vol-I-III, Churchill, 2002
- 2) Christine Evans, Genetic Counseling: a psychological approach, Cambridge University Press, 2006
- 3) Young, Introduction to Risk Calculation in Genetic Counseling, 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 Dilemmas in Genetics & genetic counseling, Oxford University Press, 2014.
- 8) McKinsey L. Goodenberg et al., Practical genetic counseling for the laboratory, Oxford, 2017.

MHG402: Clinical Genetics

**Chapter-1**

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

**Chapter-2**

- 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

**Chapter-3**

- 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

#### **Chapter-4**

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

4.2.4 Schizophrenia

#### **Chapter-5 Rare Genetic Diseases**

5.1. Introduction to rare genetic diseases

5.2. Lysosomal storage diseases

5.3. Neurofibromatosis

5.4. Progeria

5.5. Werewolfs yndrome

5.6. Skeletal dysplasia

#### **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, 4<sup>th</sup> Edition, BMJ, 2015.
- 3) Human Heredity: Principles and Issues by Micheal R. Cummings; 11<sup>th</sup> edition, Cengage Learning, 2016,.
- 4) Drs .Peter Turnpenny and Sian Ellard., Emery's Elements of Medical Genetics, 15<sup>th</sup> edition, Elsevier, 2017.
- 5) Emerys & Rimoin, Principles & Practice of Medical Genetics, 7<sup>th</sup> Edition, Elsevier, 2017.

MHG403: Human Genetic Disorders & Society-II

#### **Chapter-1 Recombinant DNA Technology**

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

#### **Chapter-2 Single Gene Disorders**

2.1. Hemophilia

2.2. Cystic Fibrosiz

2.3. Sickle cell anemia

2.4. Huntington disease

2.5. Fragile X Syndrome

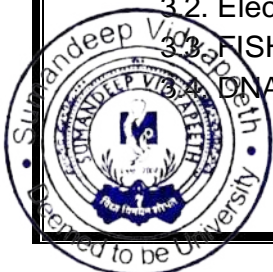
#### **Chapter-3 Molecular Genetic techniques**

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 and Enzymes used in RDT

### Chapter-4 Rare Genetic Diseases

4.1. Introduction to rare genetic diseases

4.2. Lysosomal storage diseases

4.3. Progeria

4.4. Werewolf syndrome

### Chapter-5 Genetic Counseling in Community

5.1. Genetic counseling: 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 counseling registers

5.6. Role of PC and PNDT Act, 1994 in genetics

### 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 Dilemmas in Genetics & genetic counseling, Oxford University Press, 2014.
- 4) Helen M Kingston, ABC of Clinical genetics, 4<sup>th</sup> Edition, BMJ, 2015.
- 5) Emery & Rimoin, Principles & Practice of Medical Genetics, 7<sup>th</sup> Edition, Elsevier, 2017.
- 6) Drs. Peter Turnpenny and Sian Ellard., Emery's Elements of Medical Genetics, 15<sup>th</sup> edition, Elsevier, 2017.

### MHG405 Practicals based on Theory MHG401- 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-conceptual 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.



## 7. Scheme of Evaluation

### 7.1 Semester Examination

#### 7.1.1 Internal Examination

##### Theory

Total marks: 30 (Calculated from Best of two internal exams)

Total internal examinations to be conducted: 02

##### Practical

Total marks: 20 (Calculated from Best of two internal exams)

Total internal practical examinations to be conducted: 02

#### 7.1.2 End-semester Examination

##### Theory

Total Marks: 100 (70 + 30 internal marks)

##### Practical

Total Marks: 50 (30 + 20 internal marks)

#### 7.1.3 Passing standard: 50%

## 8. Paper Pattern

### 8.1 Internal Theory Exam

Time 1.5 hours	Max. Marks 30
Question-1 Write answers to Objective type of questions (Any 05)	5x1= 05
Question-2 Write short answer questions (Any 10)	10x2= 20
Question-3 Long answer question (Any one)	1x5= 05

### 8.2 Internal Practical Exam

Time 2 hours	Max. Marks 20
Question-1 Major exercise/s	10
Question-2 Minor Exercise/s	05
Question-3 Viva-voce	05

### 8.3 End-semester Theory Exam

Time 3 hours	Max. Marks 70
Question-1 Write answers to Objective type of questions (Any 10)	10x1= 10
Question-2 Write short answer questions (Any 10)	10x5= 50
Question-3 Long answer question (Any one)	1x10= 10

### 8.4 End-semester Practical Exam

Time 3 hours	Max. Marks 30
Question-1 Major exercise/s	15
Question-2 Minor Exercise/s	05
Question-3 Viva-voce (with dissertation presentation in semester-IV)	10

## 9. Project/ Dissertation work

Project work will be carried out individually and mandatorily.

Projects will be allotted by the end of semester II.



- c. In-house projects are preferred.
- d. Students may be allowed to carry out the project work in collaboration with other research institutes, start-ups or larger companies.
- e. Co-guides from the collaborating institution/company are allowed.
- f. Internal assessment on project work will be made by the guide/s for 30 marks and will be based on the student's day to day performance in the laboratory.
- g. The dissertation will be evaluated by two external examiners.
- h. The project viva voce examination will be held at the end of last semester. The mark for the same will be added in the practical.



**Attested CTC**

*Sharaney*  
15/2/2021

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**Sumandeep Vidyapeeth**  
**An Institution Deemed to be University**  
**Vill. Piparia, Taluka: Waghodia.**  
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