# B. Sc. 6<sup>th</sup> SEMESTER DISCIPLINE SPECIFIC ELECTIVES (DSEs)

**HG616D: HUMAN GENETICS** 

**CREDITS: THEORY 4, PRACTICAL 2** 

# **OPTION - I**

## HG616DA: HUMAN GENETICS - CLINICAL GENETICS/ MEDICAL GENETICS

## **THEORY SYLLABUS**

# **UNIT 1: Replication Errors and their Repair**

- 1.1 DNA damage (by deamination, alkylation, radiation and base analogues)
- 1.2 DNA Repair
- 1.3 Diseases caused by defect in DNA repair (Xeroderma pigmentosum, Bloom syndrome, Cockayne syndrome)
- 1.4 Southern blotting and its application in disease diagnosis

# **UNIT 2: Genetics of Carcinogenesis**

- 2.1 Genetic and environmental basis of carcinogenesis
- 2.2 Tumor suppressor genes and oncogenes
- 2.3 Retrovirus and their role in cancer
- 2.4 Colon cancer and Familial melanoma

# **UNIT 3: Genetics of Immune system**

- 3.1 Innate and adaptive immune system (Basic concept)
- 3.2 Major classes of immunoglobulins and genes associated with them
- 3.3 Major Histocompatibility complex
- 3.4 Immunodeficiency diseases (SCID, X linked agammaglobulinemia, leucocyte adhesion deficiency)

# **UNIT 4: Population Genetics**

- 4.1 Principles for screening genetic disease in population
- 4.2 New born screening (like PKU, galactosemia, hyperthyroidism) and their effective interventions
- 4.3 Prenatal diagnosis of Genetic diseases (amniocentesis, chorionic villus sampling and ultrasonography)
- 4.4 Gene therapy by retroviral vector

### PRACTICALS (2 CREDITS)

- 1. ABO blood grouping
- 2. Study of Barr body from smear of buccal epithelial cells
- 3. Identification of structural and numerical aberrations through karyotype
- 4. Project work (PTC tasting/Albinism/twin study/ any genetic trait in population)

# B. Sc. 6<sup>th</sup> SEMESTER DISCIPLINE SPECIFIC ELECTIVES (DSEs)

**HG616D: HUMAN GENETICS** 

**CREDITS: THEORY 4, PRACTICAL 2** 

### **OPTION - II**

**HG616DB: HUMAN GENETICS - REPRODUCTIVE GENETICS** 

## **THEORY SYLLABUS**

# **UNIT I: Male Reproductive System**

- 1.1 Anatomy of Male reproductive system.
- 1.2 Male sex hormones
- 1.3 Spermatogenesis
- 1.4 Hormonal control of reproduction

# **Unit II. Female Reproductive System**

- 2.1 Anatomy of female reproductive system
- 2.2. Female sex hormones
- 2.3 Oogenesis
- 2.4 Hormonal control of oogenesis

# **Unit III. Female reproductive Cycle and Pregnancy**

- 3.1. Menstrual Cycle
- 3.2 Hormonal control of menstrual cycle
- 3.3 Fertilization
- 3.4. Embryonic development upto three germ layers

### **UNIT III: Reproductive Genetics**

- 4.1 Genetics of sex determination & sexual differentiation
- 4.2 Reproductive technologies, artificial insemination, cryo-preservation of oocyte, sperm & embryo
- 4.3 In vitro fertilization, embryo transfer, intra-cytoplasmic sperm injection, ethical issues, prenatal diagnosis, pre-implantation genetic diagnosis (PGD)
- 4.4 Genetic technologies used in PGD, Genetic causes of male and female infertility,

# PRACTICALS (2 CREDITS)

Study of Histological studies

- 1. Male Reproductive System
  - a) T.S. Testes, T.S. Sperm Structure, C.S. Penis
  - b) Spermatogenesis
- 2. Female Reproductive System
  - a) structure of mammalian Ovary
  - b) Oogenesis in Mammals
- 3. Study of Embryo upto three germ layers in mammals (various stages)

# B. Sc. 6<sup>th</sup> SEMESTER DISCIPLINE SPECIFIC ELECTIVES (DSEs)

**HG616D: HUMAN GENETICS** 

**CREDITS: THEORY 4, PRACTICAL 2** 

## **OPTION - III**

HG616DC: HUMAN GENETICS - CELL COMMUNICATION AND SIGNALING

## **THEORY SYLLABUS**

- 1.1. Overview of extracellular and intracellular signaling.
- 1.2. Basics of cell signaling paracrine, endocrine, autocrine.
- 1.3. Secondary messengers and their role in cell communication and signaling
- 1.4. G-protein coupled receptors and Tyrosine Kinase receptors.

# Unit II: Cell cycle

- 2.1. An overview of cell cycle and Components of cell cycle control system
- 2.2. Necrosis, senescence, programmed cell death (apoptosis).
- 2.3.. Mechanism of necrosis, senescence and programmed cell death (intrinsic and extrinsic factors).
- 2. 4. Apoptosis in relation with Cancer

# **UNIT III: Chromosome Banding Techniques**

- 3.1. Chromosome nomenclature
- 3.2 Chromosome banding techniques.
- 3.2. Molecular correlates of chromosome bands and fragile sites.
- 3.3. Use of Human cyto-genetics in medical science

## **UNIT IV: Gene Mapping**

- 4.1 Genetic mapping of Mendelian characters:
- 4.2 Recombinants, Non-recombinants, Genetic markers,
- 4.3. Two point mapping, Multipoint mapping,
- 4.4 Fine mapping using extended pedigrees and ancestral haplotypes

# PRACTICALS (2 CREDITS)

- 1. Basic sterilization required for cytological techniques.
- 2. Numericals on structural and numerical aberrations
- 3. Peparation Pedigree charts
- 4. Preparation of Karyotype from Images of chromosomes
- 5. Demostration of cell culture techniques