**HUMAN GENETICS 6TH SEMESTER (CBCS)**

**DISCIPLINE SPECIFIC ELECTIVE**

**Course Title: Clinical Genetics/ Medical Genetics**

**Course Code: DSE – HG-601**

**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**

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)