**HUMAN GENETICS Semester II (CBCS)**

**Core Course Title: Cell Biology and Genetics – II Code: DSC- HG-16-201**

**UNIT I: CELL BIOLOGY**

* 1. Cell surface receptors: Classification and Structure
  2. Signaling through G-Protein Coupled receptors
  3. Signal transduction, Second messenger system
  4. Tyrosine kinase linked receptors, JAK-STAT pathway.

**UNIT II: QUANTITATIVE INHERITANCE**

2.1. Difference between Monogenic and Polygenic traits

2.2. Polygenic inheritance of continuous traits

2.3. Genetic susceptibility in multifactorial disorder (Alcholism, Diabetes, Obesity)

2.4. Epigenetics: Effects of Environment on gene expression

**UNIT III: CHROMOSOMAL ABNORMALITIES**

3.1**.** Structural chromosome abnormalities: Cri- du- chat syndrome, Chronic myelgenous leukemia, Wolf Hirschhord syndrome.

3.2. Autosomal chromosome abnormalities: Euploidy/ Aneuploidy, Down syndrome, Patau syndrome, Edward syndrome

3.3. Sex chromosomal abnormalities: Klinefelter syndrome, Turner syndrome, Supermale and

Superfemale syndrome.

**UNIT IV: CLINICAL GENETICS**

4.1**.** Monogenic diseases with well known pathway; Cystic fibrosis, Tay-sachs syndrome, Colour blindness.

4.2. Inborn errors of metabolism and their genetic basis; Phenylketonuria, Alkptonuria.

4.3. Neuro genetic disorders: Alzheimer’s disease, Huntington disease,

Muscle genetic disorder- Duchene muscular dystrophy.

4.4. Haemophilic disorders: Sickle cell anaemia, Thalesemia, Haemophilia

**LABORATORY WORK**

1. Numericals problems on Mendelian and Non- Mendelian F2 ratios
2. Complementary gene interactions
3. Inhibitory gene interactions
4. Duplicate gene interactions
5. Supplementary gene interactions
6. Lethal gene interactions
7. PTC tasting
8. Karyotype studies of Humans, Preparation techniques of Idiograms.
9. Culture of Drosophila (Techniques)
10. Examination of different variants in Drosophila.
11. Survey of Local Area for different genetic diseases.

**Books Recommended**

1. Nelson, D.L. and Cox, M.M. Lehninger's Principles of Biochemistry. W.H. Freeman and Co., New York, 5th ed.
2. Voet, D., Voet, J.G. and Pratt, C.W. Fundamentals of Biochemistry: Life at the Molecular Level. John Wiley and Sons, New York
3. Biochemistry, Stryer
4. Molecular Biology of Cell, Bruce Alberts
5. Lodish et al: Molecular Cell Biology
6. Principles of Genetics , D. Peter Sinustad & Michael J Simmon ,Wiley & Sons, Inc
7. Dobzhansky: Genetics and origin of species
8. Brooker, R.J. Genetics, Principles and Analysis. Addison Wesley Longman, California. 2. Cummings, M.R. (2009). Human Genetics. Cenage Learning, USA.
9. Gardner, E.J. Human Genetics. Viva Books Pvt. Ltd., India.
10. Klug, W.S. and Cummings, M.R. Concepts of Genetics. Pearson Education, Inc., New Delhi.
11. Snustad, D.P. and Simmons, M.J. Principles of Genetics. John Wiley and Sons, Inc., New York.
12. Strickberger, M.W. Genetics. Prentice-Hall India Pvt. Ltd., New Delhi
13. . Medical Genetics; Jorde, B; Carey, C; Bamshad; Elsevier Publication
14. Genetics; Elrod, Stansfield; Tata Mc Graw Hill.
15. Human Genetics; Lewis 11th Edition, McGraw Hill
16. Genetics: L. Hartwell et al, McGraw Hill.