Government Degree College Baramulla

SEMESTER 8th **MAJOR COURSE**

Subject: Clinical Biochemistry

Title: Medical Genetics **Code: CBCC2822M**

CREDIT: (4+2) THEORY: 04; PRACTICAL: 02 **Contact Hours: 64(T) + 64 (L)**

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Course Objectives

- *To understand the fundamental principles of genetics and chemical basis of heredity*
- *To learn the role of genetics in medical practice*
- *To explain the role of genetic variation and mutations in human health and disease*
- *To introduce the concept of epigenetics as a key mechanism of regulation of gene expression*
- *To analyze the role of epigenetics modifications in diseases in health and disease*

Learning outcomes: At the end of the course students will be able to:

- *Identify key genetic principles and their relevance to human disease*
- *Know about the prevalent heredity diseases*
- *Recognize that epigenetic mechanisms affect gene expression and epigenetic problems can result in human diseases including cancer.*
- *Develop critical thinking in assessing genetic and epigenetic research in clinical practice*

Unit I: Introduction to Genetics: (16 Hours)

Mendel and his experiments. Laws of inheritance. Chromosomal basis of segregation and independent assortment. Test and back cross, incomplete dominance, co-dominance, pleitropy, multiple allelism, polygenic inheritance, epistasis, penetrance and expressivity with examples. Extrachromosomal inheritance & importance

Unit II: Genetics in Medical Practice -I (16 Hours)

Genetic principles and their application in medical practice; Case studies (Interacting with patients, learning family history and drawing pedigree chart); Pedigrees- gathering family history, pedigree symbols, construction of pedigrees; Monogenic traits - Autosomal inheritance-dominant and recessive; Sex-linked inheritance- dominant and recessive; Sex-limited and sex-influenced traits; Y-linked

Unit III: Genetics in Medical Practice -II (16 Hours)

Syndromes and disorders: Definition and their genetic basis. Types of mutation, mutagens - physical and chemical, detection of mutations. Structural and numerical aberration of chromosomes. Hereditary defects–(Klinefelter, Turner, Cri-du-chat, and Downs syndromes, Cystic fibrosis and Tay Sach's Syndrome).

Unit IV: Epigenetics in health and disease (16 Hours)

Chromatin structure, Histone modifications and their roles in gene expression, chromatin-remodelling complexes, Histone modifications in cancer & metabolic diseases. DNA methylation. Significance of hypomethylation and hypermethylation, Regulation of eukaryotic gene expression by long non-coding RNAs, and miRNAs, Aberrant DNA methylations in cancer& metabolic diseases.

Practicals (2 credits: 64 Hours)

1. Extraction of DNA from body fluids.

2. Karyotyping with the help of photographs/teaching kits.
3. To study the karyotyping of chromosomes from the given animal samples
4. Identification of inactivated X chromosomes as barr body from the given sample
5. Methylation specific-PCR to differentiate and detect unmethylated versus methylated cytosines.

Books recommended:

1. Principles of Genetics by Gardner, E.J., Simmons, M.J., Snustad, D.P. Wiley India.
2. Principles of Genetics by Snustad, D.P., Simmons, M.J. V Edition. John Wiley and Sons Inc.
3. Concepts of Genetics by Klug, W.S., Cummings, M.R., Spencer, C.A. Benjamin Cummings.
4. Epigenetics by Allis, Jenuwein, and Reinberg. Cold Spring Harbor Press.
5. Epigenetics by Lyle Armstrong. Garland Science.
6. The Epigenetics Revolution: by Nessa Carey. Columbia University Press