

# THE NATIONAL COLLEGE BASAVANGUDI, BANGALORE-560004 [AUTONOMOUS] DEPARTMENT OF ZOOLOGY

# Exploring Genetic Variations: Clinical Applications in Molecular Genetics

**Duration: 30 Hours** 

Course code: ZO-CMG

## **Course Description:**

Exploring Genetic Variations: Clinical Applications in Molecular Genetics is designed to provide students with a comprehensive understanding of the principles, techniques, and applications of molecular genetics in clinical settings. Through theoretical lectures, laboratory exercises, case studies, and research projects, students will explore various genetic variations, their implications for human health, and the role of molecular genetics in disease diagnosis, prognosis, and treatment. Emphasis will be placed on practical skills development, ethical considerations, and emerging trends in clinical molecular genetics.

## **Course Objectives:**

- 1. Understand the fundamentals of molecular genetics and its relevance to clinical practice.
- 2. Gain proficiency in laboratory techniques used for genetic analysis, including DNA sequencing, PCR, and gene expression analysis.
- 3. Learn about common genetic variations, including single nucleotide polymorphisms (SNPs), insertions, deletions, and chromosomal aberrations.
- 4. Explore the role of molecular genetics in disease diagnosis, risk assessment, and personalized medicine.
- 5. Develop skills in interpreting genetic test results and communicating findings to patients and healthcare professionals.
- 6. Understand ethical, legal, and social implications (ELSI) of clinical molecular genetics research and practice.
- 7. Stay abreast of emerging technologies and advancements in the field of clinical molecular genetics.

# Module 1: 7 Hours

#### Introduction to Clinical Molecular Genetics

- Definition and scope of clinical molecular genetics
- Historical perspective and milestones in the field
- Applications of molecular genetics in clinical practice

# Laboratory Techniques in Molecular Genetics

- DNA extraction and purification methods
- Polymerase Chain Reaction (PCR) and its variants
- DNA sequencing techniques (Sanger sequencing, Next-Generation Sequencing)



Module 2. 7 Hours

Genetic Variation and Human Health

- Types of genetic variations (SNPs, insertions, deletions, copy number variations)
- Relationship between genetic variations and human diseases
- Genomic instability and chromosomal aberrations

Genetic Testing and Diagnostic Techniques

- Overview of genetic testing methods (molecular, cytogenetic, biochemical)
- Preimplantation genetic diagnosis (PGD) and prenatal screening
- Cancer genetic testing and hereditary cancer syndromes

Module 3. 8 Hours

Pharmacogenetics and Personalized Medicine

- Role of genetic variations in drug response and metabolism
- Pharmacogenetic testing and drug dosing optimization
- Implementation of personalized medicine approaches in clinical practice

Gene Expression Analysis and Functional Genomics

- Gene expression profiling techniques (microarrays, RNA sequencing)
- Functional annotation of genetic variants
- Systems biology approaches to understanding disease mechanisms

Module 4: 8 Hours

Interpretation of Genetic Test Results

- Variant classification and interpretation guidelines
- Reporting genetic test results to patients and healthcare providers
- Genetic counselling and patient education

Ethical, Legal, and Social Implications (ELSI)

- Ethical considerations in genetic testing and research
- Genetic privacy, consent, and confidentiality
- Societal impact of genetic testing and implications for healthcare policy

#### Recommended Textbooks:

- "Principles of Clinical Molecular Genetics" by H. Helen Hui and J. Milburn Jessup
- "Genetics in Medicine" by James S. Thompson and Margaret W. Thompson
- "Clinical Genomics: Practical Applications in Adult Patient Care" by Michael T.
   Gerhardt and William T. Kearns

