YENEPOYA
(DEEMED TO BE UNIVERSITY)
Recognized under Sec 3(A) of the UGC Act 1956

Genetics and Genomics

Core course for Pre-PhD: 4 credits

Yenepoya Research Centre
Yenepoya (Deemed to be University)
University Road, Deralakatte
Mangalore – 575018

Course Name- Genetics and Genomics

1.	Course Type	:	Core
2.	Level	:	Ph.D. (Pre-PhD course work)
3.	Credit Value	:	4 Credits
4.	Total Hours	:	60 (L:P:S: 10:25:25)
5.	Total Marks:	:	100 (IA= 40 + Final exam= 60)

6. Course Objectives:

- Provide knowledge of the fundaments of genetics, the central dogma, key molecules and applications of molecular biology in genetics.
- Sensitize students to understand Mendelian genetics, the human genome, importance of population genetics, common genetic diseases and importance of diagnostic genetics.
- Expose research scholars to different laboratory skills used in genetics research and train research scholars to apply the use of genome sequencing and data analytical tools to solve research problems.

7. Learning Outcomes

- Know, understand and appreciate the importance of classical and human genetics.
- Know and understand genome, its organization and study human genetic diseases and apply the knowledge of genetic engineering
- Understand the principles of gene manipulations and its applications and apply the analytical methods for genetics research, cell and molecular biology techniques.

8. Competencies

1. Describe the functions of the different biological macromolecules in cellular communication and cell cycle regulation.
2. Define the principles of Mendel's laws of inheritance, its exceptions and correlate different aspects of population genetics.
3. Demonstrate the use of the important analytical experimental tools and equipments used for genomic research
4. Categorize the congenital and common genetic diseases based on the mechanism of genetic alterations.
5. Make use of the data generated from the Human genome project for studying genetic variations
6. Comprehend the use of genome sequencing in research and clinical diagnosis.
7. Perform experimental procedures required for gene expression assays, mutation studies and recombinant DNA technology.
8. Discuss bioethical issues arising from recombinant DNA technology, genetically

modified organisms (GMO) and importance of genetic counseling.

9. Practice of care and safety protocols in handling chemicals, biohazardous materials or equipments and disposal of chemicals biological/hazardous wastes as per prescribed guidelines.

Contents of the Course

Module I: Bio Analytical Methods (12 h)

1.1. Microscopy- Image formation, resolution, Types (Light, Fluorescence, Confocal, SEM and TEM) techniques and applications, UV-Vis, Fluorescence Spectroscopy

1.2. Separation Methods-Centrifugation-Sedimentation principle, differential centrifugation, different types of centrifuges, Chromatography- types (adsorption, partition, affinity, ion exchange and size exclusion) and applications

1.3. Biological techniques-PCR, qRT-PCR, Electrophoresis, blotting techniques, ELISA, Cell biology techniques: Assessment of cellular viability, cell proliferation and migration assays, Histopathology and immunocytochemistry, Flowcytometry

1.4. Physical and chemical methods of sterilization, Media and buffers, Mammalian and Microbial culture techniques

Module 2: Cell and Molecular Biology (12 h)

2.1. Functions of biological macromolecules – lipids, proteins and nucleic acids– structure and function, Applications of Central Dogma, Viral, Prokaryotic and eukaryotic cells and their genomes.

2.2. Importance of Cell cycle regulation- cellular differentiation, proliferation, apoptosis

2.3. Basic principles of Pharmacology/toxicology– Agonist, Antagonist, Receptors, Dose response relationships, Cellular communication, signaling mediated transport of ions and molecules across cell membrane

2.4. Uses of Enzymes- DNA polymerase, restriction endonucleases, reverse transcriptase, kinase, Cloning vector (characteristics applications) Plasmids Vectors, Gene cloning, Cloning Strategies

Module 3: Classical and human genetics (12 h)

3.1. Cell cycle & Mitosis and Meiosis; Mendel's laws and exceptions, Multiple alleles; Dominance relations; Importance of Pedigree charts; Autosomal dominant & recessive inheritance; X-linked dominant & recessive inheritance

3.2. Importance of Karyotypes; Variations in Chromosome number and structure; Sex chromosome aneuploidy; Dosage compensation; Sex linked inheritance, Linkage and Linkage Disequilibrium; Mitosis, Meiosis and Crossing over

3.3. Functionality of the Human Genome Project; Polygenic traits and inheritance; Multifactorial Traits examples. Single Nucleotide polymorphisms, GWAS, Ribosomal sequencing for molecular taxonomy.

Module 4: Genome and Genetic diseases (12 h)

4.1. Importance of genome organization: eukaryotic and bacterial chromosomes and cellular components, histones and nucleosomes, cell cycle and checkpoints, genome size, gene dosage, repetitive DNA, mobile DNA elements

4.2. Understand congenital and common genetic diseases, prevalence and databases. Common syndromes due to numerical chromosome changes, Common syndromes due to structural alterations (translocations, duplications, deletions, microdeletion, fragilesites). Disease conditions (autosomal dominant, autosomal recessive, X- linked dominant, X-linked recessive and complex disease conditions with examples)

4.3. General concept of a gene, Operon, Non-coding genes. C-value paradox. Variation at the genetic level: DNA markers-VNTR, STR, microsatellite, Mutation detection SNP and their detection techniques-RFLP, Single gene and multiple gene disorders.

Module 5: Genetic Engineering (12 h)

5.1. Principle behind Sanger sequencing-, methodology and applications, Human Genome sequencing project; Analysis of gene expression- qPCR, northern blot, southern blot; exome sequencing; DNA microarrays; Copy number variation, Next Generation Sequencing (NGS) technology, Basics of sample preparation, Whole genome and exome sequencing.

5.2. Importance of Mutation (Classification, mechanism, repair, role in genetic analysis and evolution) Changes in Chromosome number and structure (Polyploidy, aneuploidy,

5.3. Chromosomal rearrangements - deletion, duplication, inversion, and translocation), Genetically modified organisms, Bioethical issues

5.4. Application of Cloning: Plasmid vectors, PCR, gene isolation by PCR, primer design – gene specific primers, types of PCR – inverse PCR, multiplex PCR, nested PCR, TOPO cloning, cloning of PCR products – TA cloning, blunt end cloning, cloning with added restriction sites.

Teaching-learning methods (module and unit wise)

Modules	**Teaching-learning**		
	Lecture	Practical/Hands on	Self-study
Module 1: Bio-Analytical Methods	1.1		
		1.2	
		1.3	
			1.4
Module 2: Cell and Molecular Biology	2.1		
		2.2 (Seminar)	
		2.3 (Team work)	
	2.4		
Module 3: Classical and Human genetics	3.1		
			3.2 (Seminar)
	3.3		
Module 4: Genome and genetic diseases			4.1 (Seminar)
	4.2		
		4.3(Team work)	
Module 5: Genetic Engineering		5.1 (Group Discussion)	
	5.2		
			5.3 (Seminar)
		5.4	

10. Assessments

Formative assessments: (40 marks)

1	Internal Exams - 40 marks each (2)	20 M
2	Seminar (2)	8 M
3	Group discussion (2 Including ethical and regulatory issues)	6 M
4	Case studies (2)	6 M

Summative Assessment: (60 marks)

Sl. no.	Details	Q X M
1	Instrumentation- Application of any of the major instruments used in the field of genetics and molecular biology for generating data (example; use of Sanger sequencing machine)	2X10M= 20 M
2	Interpretation of data (example: PCR /RT-PCR/Design a primer from given sequence/pedigree trees)	1X20M=20 M
3	Write the methodology for solving a given research problem (example: Identification of a novel gene/variant associated with a particular disease)	
4	Descriptive questions to assess the knowledge from module 2 and 3	4X5M=20 M

*A question bank will be maintained with multiple scenarios.

Learning Resources

Student should refer leading Journals and publishers in the subject category and list is not limited to specific titles.

Reference books

1. Snustad, Simmons (2017). Principles of Genetics 7th edition. John Wiley and Sons.
2. Benjamin Pierce (2016). Genetics: A Conceptual Approach, W H Freeman & Co.
3. Klug, Cummings, Spencer, Palladino (2015). Concepts of Genetics: International Edition, Pearson Education.
4. Brown T. A. (2016). Gene Cloning and DNA Analysis: An Introduction, 7th Edition. Wiley Blackwell.
5. Brown T. A. (2017). Genomes. Garland Science.
6. Peter J Russell (2016). I Genetics: A Molecular Approach, 3rd Edition. Pearson, India.

Online sources

- https://currentprotocols.onlinelibrary.wiley.com/journal/19348258
- https://www.ncbi.nlm.nih.gov/pmc/articles/PMC1801278/
- http://cshprotocols.cshlp.org/site/Taxonomy/genetics_l1.xhtml
- https://www.genome.gov/about-genomics/teaching-tools/Genomics-Education-Websites
- https://www.qmul.ac.uk/library/library-skills/resource-guides-by-subject/biological-sciences/useful-websites/genetics---useful-websites/
- https://iupui.libguides.com/genetics/MobileAppsOnline
- National Ethical Guidelines for Biomedical and Health Research involving Human Participants", 2017, published by Indian Medical Research, 2017,