Course Code: MLC401 **Course Title: Clinical Genetics I**
Number of Credits: 3T + 2P = 5
Effective from AY: 2022 -2023

Prerequisite for the Course:	Basic knowledge of cell biology and genetics.	
Objectives:	• Acquaint students with recent genetic techniques • Know about the structure and function of genetic material • Learn about structural and numerical abnormalities their inheritance pattern and pedigree analyses.	
Content:	**Module 1: Introduction to Human Genetics** Growth of human genetics; levels of genetics. Structure and composition of the human chromosome: basic structure of DNA; molecular structure and organization. Classification of Human chromosomes: Paris nomenclature / ISCN; methods of studying chromosomes; identification of individual chromosomes; Flow Karyotyping (Quantification on DNA of individual chromosomes); FACS – Fluorescence-activated cell sorter.	15hrs
	Module 2: Chromosomal Abnormalities Numerical abnormalities (somies; ploidies; mosaic; chimera; syndromes). Structural: Translocations; Deletions; Duplications; Inversion; isochromosomes; Ring chromosomes; causes for genetic abnormalities- meiotic and mitotic nondisjunction; uniparental disomy; mutations; single gene disorders.	15hrs
	Module 3: Pattern of Inheritance Autosomal Dominant, Autosomal Recessive, X-linked Dominant, X-linked Recessive, Y-linked, sex limited inheritance, sex influenced inheritance, X inactivation, Multifactorial inheritance, mitochondrial inheritance, imprinting. Pedigree analysis of some genetic disorders: Haemophilia, Color blindness, Duchenne Muscular Dystrophy (DMD), achondroplasia and PKU.	15hrs
	Practical Module : • Specimen procurement and logging for cytogenetic procedure. • Culture media preparation • Identification of Chromosomes. • Inoculation of Lymphocyte culture/peripheral blood culture. • Harvesting of Lymphocyte culture to obtain metaphase plates. • Chromosomal banding technique: GTG Banding. • Karyotyping of Human chromosomes:	4 hrs 4 hrs 4 hrs 4 hrs 4 hrs 4 hrs 4 +4 +4 hrs

	○ use of Cytovision/any other Karyotyping software ○ Microphotography ○ Image capturing, image processing, and analysis • Study of Karyotypes: Normal male and female and various syndromes • Construction of Pedigree from given data. • Analysis of pedigree charts to determine the mode of inheritance	4+ 4 hrs 4+4 hrs 4+ 4hrs
Pedagogy:	Lectures/tutorials/assignments/ Presentations/Practicals/ demonstrations.	
Learning Outcome:	By the end of this course, students will be able to 1. Understand the functions of the genetic material. 2. Correlate genetic mutations to diseases in human population. 3. Perform Karyotyping using software. 4. Construct and analyse human pedigrees.	
References	1. Jorde L, Carey J and Bamshad M(2016). Medical Genetics. Fifth edition. Elsevier Publication imprint. eBook ISBN: 9780323391979. 2. Singh BD (2014): Fundamentals of Genetics. Second Edition, Kalyani Publishers, New Delhi. 3. Matheiesen A and Roy K(2018). Foundation of Perinatal Genetic counseling. eISBN: 9780190681111 4. Gardner EJ, Simmons MJ and Snustad DP (2013): Principles of Genetics, Eighth Edition, John Wiley Publication, Singapore 5. De Robertis EDP, De Robertis EMF (2012): Cell and Molecular Biology, Eigth Edition. Wolter Kluwer Publication, Philadelphia. 6. Thompson JS, Thompson MW(1966): Thompson & Thompson Genetics in Medicine,Elsevier Publication, Philadelphia. REFERENCE BOOKS FOR PRACTICALS: 1. Arumuga N, MeyyanRP (2016): Advances in Genetics Volume 1(Dr. N. Arumugam, R P Meyyan, Saras Publication, Nagercoil, Tamil Nadu. 2. GardnerA and Davies T(2010) Human Genetics 2nd Edition,Viva books publication,Delhi.	