

Course Code: MLE401
Number of Credits: 3T + 2P = 5
Effective from AY: 2022 -2023

Course Title: Clinical Genetics II

Prerequisite for the Course:	Basic knowledge of Cell biology and genetics	
Objectives:	- To get acquainted with recent procedures used in artificial reproductive techniques and their acceptance in the society. - Techniques for analysis of samples for success of procedures conducted. - Knowledge of recent techniques used for better results and treatment. - To learn about genetic counseling and steps to help guide patient for particular medical treatment available.	
Content:	Module 1: Molecular genetics, Genetics of Cancer, Dermatoglyphics Molecular genetic techniques used in genetic diagnosis: Blotting techniques – Southern, Northern and Western, PCR/ RFLP, FISH, DNA sequencing & DNA fingerprinting. Genetics of Cancer: introduction, characteristics of cancer cells, origin of cancer cells, genes associated with cancer, environmental causes of cancer, human genome data tailor diagnosis and treatment. Dermatoglyphics: Introduction, classification, Flexion creases. Dermatoglyphics in clinical disorders, Clinical application & its advantages and limitations.	15hrs
	Module 2: Reproductive technologies, Genetics and Society Reproductive technologies: infertility and subfertility, assisted reproductive technologies (IUI, surrogate motherhood, IVF, GZIT, ZIFT), preimplantation genetic diagnosis. Genetics and Society : (i) Human genome project : (ii) Forensic science (iii) DNA finger printing application (iv) Gene therapy (v) Eugenics. vi) Stem cell research.	15hrs
	Module 3: Prenatal Diagnosis, Genetic Counselling Prenatal Diagnosis: Definition: Various procedures - Amniocentesis, Chorionic villus sampling, Ultrasonography and Fetoscopy. Genetic Counselling (Stage1: History and Pedigree Construction, Stage 2: Examination, Stage 3: Diagnosis, Stage 4: Counselling; and Stage 5: Follow up).	15hrs
	Practical Module: - Introduction to molecular genetic lab: general rules, handling of chemicals, equipments and biological materials; waste disposal. - Isolation of DNA from human blood. - Determination of the molecular size of DNA. - Analysis of DNA fingerprints and FISH images	30 hrs x 2

	• Dermatoglyphics: Recording of print of fingertips and palm. • Manual DNA sequencing and data analysis. • Amniotic fluid culture: Flask method and Coverslip method. • Chorionic villi culture: Short-term culture • Chromosomal analysis from the product of conception (abortus study) (03 Practicals) • Disease suspicion by spot tests: Fanconi's syndrome, PKU, maple syrup urine disease, Tryptophanuria.	
Pedagogy:	Lectures/tutorials/assignments/ Presentations/Practicals/ demonstrations.	
Learning Outcome:	By the end of this course, students will be able to 1. Describe and explain the molecular genetic techniques used in genetic diagnosis and reproductive techniques which can be recommended to overcome infertility. 2. Demonstrate the application of dermatoglyphic prints in disease detection. 3. Perform procedures of DNA isolation, Molecular size determination, and disease detection for inborn errors of metabolism. 4. Analyze FISH images and DNA fingerprints.	
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: 978019068111. 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, Eighth 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, Vivabooks publication, Delhi.	