

SEMESTER I

Paper DLTC 01: Clinical Genetics I:

THEORY

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 organisation.
- 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.

Module 2:

- Chromosomal Abnormalities:
- Numerical abnormalities (somes; ploidy; 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.

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.

PRACTICALS

- 1) Specimen procurement and logging for cytogenetic procedure.
- 2) Culture media preparation

4) Identification of Chromosomes.

5) Inoculation of Lymphocyte culture/peripheral blood culture.

6) Harvesting of Lymphocyte culture to obtain metaphase plates.

7) Chromosomal banding technique: GTG Banding.

8) Karyotyping of Human chromosomes (use of Cytovision / any other Karyotyping software is optional: for image capturing, image processing, and analysis).

9) Study of Karyotypes: Normal male and female.

10) Construction of Pedigree from given data.

11) Analysis of pedigree charts to determine the mode of inheritance.

Paper DLTC 02: Clinical Biochemistry I

THEORY

Module 1: ↗

- The scope of biochemistry:
- Chemical organization of the cell.
- Organic and inorganic components of the cell.
- Marker enzymes of the cell.
- Hydrogen ion concentration and buffers: pH
- Blood buffers, regulation of blood pH.
- Acid base metabolism.

} Zoology

Module 2:

- Carbohydrate chemistry.
- Protein chemistry.
- Lipid chemistry.

Module 3:

- Enzymes:- Definition, classification, factors affecting enzyme action.
- Enzyme inhibition,