

**CORE COURSE - XI – CLINICAL BIOCHEMISTRY**

**Unit 1**

Patterns of inheritance– autosomal and sex – linked disorders.

Disorders of amino acid metabolism– amino aciduria, Phenylketonuria, Hartnup disease, alkaptonuria, albinism, cystinuria, cystinosis, homocystinuria and maple syrup urine disease. Disorders of carbohydrate metabolism– glycogen storage diseases, galactosemia, fructose intolerance and fructosuria. Disorders of purine pyrimidine metabolism: Hyperuricemia and gout. Hypouricemia. Orotic aciduria. Serology: C reactive protein test, Rheumatoid arthritis (RA) test, immunologic test for pregnancy. Amniotic fluid: Origin, composition and analysis of amniotic fluid.

**Unit 2**

Blood sugar homeostasis: Role of tissues and hormones in the maintenance of blood sugar. Hypoglycemia, hyperglycemia, glycosuria. Diabetes mellitus – classification, metabolic abnormalities, diagnosis and management. Acute complications – diabetic ketoacidosis hyperosmolal non-ketotic coma. Long-term complications – retinopathy, neuropathy and nephropathy, glycosylation. Disorders of lipid metabolism – lipoproteinaemias. Lipid storage diseases – Gaucher's, TaySach's Niemann Pick and Sandhoff's disease. Fatty liver. Atherosclerosis.

**Unit 3**

Jaundice: Retention and regurgitation jaundice. Causes, consequences and biochemical findings in hepatitis and cirrhosis. Tests related to excretory (bile pigments) synthetic (plasma proteins, prothrombin time) detoxifying (hippuric acid, NH<sub>3</sub>, aminopyrine) and metabolic (galactose) functions. Gallstones. Gastric function tests: Stimulation tests – insulin and pentagastrin. Peptic ulcer, gastritis and Zollinger Ellison syndrome. Porphyrrias. Free radicals and disease: Formation of free radicals, lipid peroxidation and consequences. Antioxidant defence mechanisms.

**Unit 4**

Kidney function: Biochemical findings in glomerulonephritis, renal failure and nephritic syndrome. Nephrolithiasis. Glomerular function tests – inulin, urea and creatinine clearance tests, renal plasma flow, plasma  $\alpha_2$ -microglobulin, urea and creatinine. Tubular function tests – water load, concentration and acid excretion tests. Abnormal constituents of urine.

Clinical enzymology: Serum enzymes and isoenzymes in health and disease – Transaminases (AST, ALT) acid and alkaline phosphatases, amylase, LD and CK. Enzyme patterns in disease – Liver and muscle disease, myocardial infarction.

## **Unit 5**

Oncology: Cancer cell – morphology and growth characteristics. Biochemical changes in tumor cells. Differences between benign and malignant tumors. Tumor markers – AFP, CEA and Hcg only. Agents causing cancer – radiation, viruses, chemicals (brief account only). Multistep carcinogenesis – initiation, promotion, progression. Oncogenes and proto-oncogenes – mechanisms of proto-oncogene activation. Tumor suppressor genes – p53.

### **Books recommended**

1. Clinical Chemistry in diagnosis and treatment Mayne ELBS.
2. Clinical Chemistry Marshall 3rd edition Mosby.
3. TietZ textbook of Clinical Chemistry – 1998 3rd edition Saunders.
4. Principles of Internal Medicine. Harrison's Vol 1 & 2, 14th edition Mc Graw Hill.
5. Biochemistry and disease. Cohn and Roth. 1996, Williams and Wilkins.
6. Harper's Biochemistry McGraw Hill, 2000.
7. Biochemistry – A case oriented approach. Montgomery et al. Mosby.
8. Clinical Chemistry – Principles, procedures, correlations – Bishop, Lippincott, 2000.