Subject Code : N3O9

BIOCHEMICAL GENETICS AND CLINICAL BIOCHEMISTRY.

UNIT – I

Mutations affecting structure and synthesis of proteins:

a. Haemoglobin variants : sickle cell disease – hereditary methemogloinemias.
b. Hepatoglobin variants : Bh Freiburg Hb Harlam.
c. Thalassemias hereditary persistence of fetal haemoglobin
d. 1 – antitrypsin deficiency.

Genetic disorders such as cystic fibrosis; Duchenne muscular dystrophy and Becker's dystrophy.

UNIT – II

Inborn errors of metabolism:

a. Inborn errors of amino acid metabolism: Phenylketonuria, tyrosinosis, alkaptonuria, maple syrup urine disease, homocystinuria and histidinuria, albinism, Parkinson's disease, Diseases involving lysine and ornithine, folic acid deficiency, clinical problems related to glutathione.
b. Disorders of purine metabolism: Lesch Nyhan, Immunodeficiency diseases associated with defects in purine nucleotide metabolism, gout.
c. Galactosemia diabetes mellitus, lactic acidocis, G-6-PD defiency, mucopolysacchradoses.
d. Glycogen storage diseases: Von Gierk disease, Pompes disease, Forbes disease, Cori's disease, Mc Aldles disease.
e. Apolipoproteinuria and genetic abnormalities in lipid energy transport : Tay – Sach's disease, Gaucher's disease.

UNIT – III

Collection, preservation of biological specimen such as blood enzymes, urine, CSF, bile etc.

Biochemical assessment of liver function, pigments, bile acid metabolism, cirrhosis, hepatitis, jaundice coma, tumors and inherited abnormalities to bilirubin metabolism, Gilbert's, Crigler Najjar, Dubbin – Johnson and Rotor Syndrome.

UNIT – IV

Disorders in blood: Agranulocytosis, thrombocytopenia, aplastic, hemolytic, met – Hb anemias, heamaturia, disorders of clotting mechanisms.

Hypertension, hypercholesterolemia, LDL, dyslipidemia – an clinical diagnosis.

UNIT – V

Quantitative analysis of urine, urinary sediments, renal function tests, osmolarity and free water clearance, acute and chronic renal failure, glomerular nephritis, nephritic syndrome, renal hypertension and urinary calculi. Analysis of stones, peritoneal and haemo dialysis.

Concept of accuracy, precision, reliability, reproducibility and other factors of quality control, normal values. Definition of LD 50, ED 50. Therapeutic index, acute and chronic toxicity.

References:

1. Text Book of Biochemistry with clinical correlation : TM Devlin (3rd Edn.,), 1994.
2. Principles and practice of Medical Genetics (Vol.I) : AEH Emergy and DL.Rimoin Churchill Livingstone, 1983, chap 99.
3. Genetic Biochemical Disorders : PF Benson and AH Fensom, Oxford Monograph in Medical Genetics, 1985.
4. Practical Clinical Biochemistry : H Varley.
5. Clinical Biochemistry : W Hoffiman
6. Clinical Chemistry in diagnosis and treatement : Zilva.
7. Principles and practice of medicine : Davidson.