

COURSE I : INHERITED DISEASES AND GENETIC COUNSELLING

Unit I

Introduction : Basic principles of inheritance – chromosomes and genes – human chromosomes – karyotype – sex determination in man.

Unit II

Blood groups in man : Inheritance of ABO blood groups – MN blood group – Rh factor – ABO incompatibilities – Inherited diseases associated with blood – haemolytic anemia – Thalassemia and Cooley's anemia- Genetic counseling.

Unit III

Inborn errors of metabolism : amino acid metabolism – phenylketonuria, alkaptonuria, albinism, and genetic goitrous cretinism – sickle cell anemia – diabetes mellitus. Genetic counseling.

Unit IV

Sex linked inheritance : X-linked inheritance – haemophilia and colour blindness; Y-linked inheritance – ichthyosis hystrix; Y chromosome based problems in sex determinations and differentiations (introduction only). XY – linked genes – total colour blindness, xeroderma pigmentosum and retinitis pigmentosa.

Abnormal human karyotypes – autosomal abnormalities in man – Down syndrome (21st trisomy), Turner syndrome, Klinefelter syndrome and multiple sex chromosome syndrome. Genetic counselling.

Unit V

Inherited diseases in man : Dominantly inherited disease – Glaucoma, Alzheimer's diseases and manic depression. Recessively inherited diseases – retinoblastoma and haemolytic anemia.

Diagnosis of genetic disorders : prenatal screening – non-invasive types – ultrasonography; invasive testing methods – foetoscopy, amniocentesis and chorionic villi biopsy.

Management of genetic disorders-gene therapy

Reference:

1. Strikberger, M.W. (1985). Genetics, Prentice Hall of India, New Delhi.
2. Stine, C.J. (1989). The new human genetics. Wm.C.Brown Publishers, Iowa.
3. Sarin, C.Genetics (1985). Tata Mcgraw Hill publishing Co., New Delhi.
4. Verma, P.s. and Agarwal, V.K.(1998). Concept of genetics, human genetics and eugenics, S.Chand & Co., Ltd, New Delhi.