



National University of Sciences and Technology

Course Description

Course Title	Course Code	Credit Hours
Medical Genetics	MVI-851	3 – 0

TEXT BOOK:

- ABC of Clinical Genetics (3rd Edition), Helen M Kingston, BMJ Books, London WC1H 9JR, 2002

REFERENCE BOOK

- Medical Genetics at a Glance, DORIAN J. PRITCHARD and BRUCE R. KORF, (3rd Edition), BlackWell Science 2013 (latest edition).
- LEWINS GENES X, Jocylyn EK, Elliott SG, Stephen TK, Jones and Bartlett Publishers 2010

COURSE OBJECTIVES:

- Outline what genetic counseling is, why and when it is necessary to give genetic counseling, and how it is regulated in different countries
- Point out central examples of monogenic, polygenic and chromosomal disorders
- Discriminate between different patterns of mendelian inheritance of monogenic diseases, and outline genetic and biochemical mechanisms of some central monogenic disorders
- Demonstrate a strong background in mechanisms underlying numerical and structural chromosomal aberrations and principles mediating chromosomal disease
- Describe main principles in immunogenetics
- Outline central principles and examples of both sporadic and hereditary cancers
- Genetics of diabetes and neurological disorders

COURSE OUTLINES

- Introduction to Genetics
- Genetic Assessment and Counseling
- DNA Replication and Recombination
- DNA Repair System
- Mendel Genetics
- Developmental and Chromosomal abnormalities
- Genetics of Cancer
- How Cells Read the Genome: From DNA to RNA
- From RNA to Protein
- Genetics of Neurological Disorders
- PHARMACOGENETICS and cystic fibrosis
- Nutrigenomics

ASSESSMENT

Description	Percentage Weightage (%)
Assignments	05-10%
Quizzes	10-15%
Mid Semester Exams	30-40%
End Semester Exam	40-50%