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| **Course title** | **Code** | **Semester** | **Type of course** | **Course volume (Contact hours)** | **ECTS** |
| **Medical Genetics I** | **MED**  **2011** | **III** | **Mandatory** | 28 | **2** |
| **Faculty, the educational program and level of education** | * School of Medicine and Health Sciences * Higher Medical Educational Program “Medicine” * One cycle 6-year | | | | |

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| **Learning Course Content** |
| * Genetics and Genomics in Medicine, challenges and promising future. Overview of human genetics research methods (Genealogical, twins, cytogenetic, ontogenetic, tissue culture etc. Genetic testing, genome sequence and use in Mutations. Types of mutations: An overview of genomes, chromosomes and point/ gene mutations. Mutagenic factors. Gene polymorphism. modern medicine. * Clinical cytogenetics; Basic methods of genome analysis (fluorescence in situ hybridization). Chromosome Identification. Clinical indications for chromosome and genome analysis. * The chromosomal and genomic basis of disease: disorders of the autosomes and sex chromosomes. Down syndrome, microdeletion and duplication syndromes, idiopathic chromosome abnormalities, Klinefelter syndrome, Turner syndrome * Single gene inheritance. Penetrance and expressivity (autosomal dominant and autosomal recessive inheritance, etc.). Pedigree analysis. Polyglutamine disorders, Flagile X syndrome, neurofibromatosis 1, Duchenne muscular dystrophy, hemophilia, Rett syndrome, Huntington disease, Achondroplasia. * Complex inheritance of common multifactorial disorders. Coronary artery disease. Type 1 diabetes mellitus, Alzheimer disease. * Developmental genetics and birth defects. Genes and environment in development. Malformation, deformation, and disruptions. Cellular and molecular mechanisms in development * Molecular bases of genetic disorders – mutation influence on protein function. Hemoglobinopathies (Thalasemias, Hemolytic disorders), expression of globine genes, dosage influence. * Genetic disorders caused by receptor proteins (familiar hypercholesterinemia etc.) and transport proteins (cystic fibrosis etc.). Genetic Disorders caused by defective enzymes (Aminoacidopathies, Phenilketonuia, Tay-sax disease, porphyria etc.) |
| **Textbooks and Materials** |
| * Thompson & Thompson genetics in medicine - Robert L. Nussbaum, Roderick R. Mc. Innes; Elsevier; 8th ed. 2016. * Concepts of Genetics-William S. Klug; Michael R. Cummings; Pearson; 12th.ed. 2019; * EMERY AND RIMOIN'S ESSENTIAL MEDICAL GENETICS - DAVID L. RIMOIN'S; ELSEVIER; 6TH ED.2013; * LEWIN'S GENES XI - KREBS, JOCELYN E; JONES AND BARLETT LEARNING; XI ED. 2014. * MOLECULAR BIOLOGY OF THE CELL- BRUCE ALBERTS, ALEXANDER JOHNSON; GARLAND SCIENCE; 6TH. ED. 2015; * HUMAN EMBRIOLOGY AND DEVELOPMENTAL BIOLOGY - BRUCE M. CARLSON; ELSEVIER SOUNDER; 5TH.ED.2014; * THE PRINCIPLES OF CLINICAL CYTOGENETICS - STEVEN L.GERSEN; MARTHA B KEAGLE. HUMANA PRESS; 2ND. ED. 2005; * HUMAN MOLECULAR GENETICS - TOM STRACHAN;ANDREW READ; GARLAND SCIENCE; 4TH.ED. 2011. * ABC OF CLINICAL GENETICS – HELEN MKINGSTON. 3 RD EDITION. 2002; |