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| **Course title** | **Code** | **Semester** | **Type of course** | **Course volume (Contact hours)** | **ECTS** |
| **Medical Genetics II** | **MED**  **2012** | **IV** | **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** |
| |  | | --- | | * Prenatal diagnostics. Prenatal diagnosis in invasive testing. Screening for neural tube defects. Screening for down syndrome and other aneuploidies. Problems during prenatal chromosomal analysis. * Mitochondrial genome. Maternal inheritance. Mitochondrial genome mutations and diseases * Cancer genetics and genomics. Genetic basis of cancer. Driver and passenger gene mutations. Activated oncogenes and tumor suppressor genes. Cellular heterogeneity within individual tumors. Cancer in families. Retinoblastoma. Familial breast cancer due to mutations in BRCA! And BRCA2. Hereditary colon cancer. Familial adenomatous polyposis. Lynch syndrome. Sporadic cancer. Cytogenetic chances in cancer. Gene expression profiling in cancer prognosis. Cancer and environment * Epigenetics and gene expression characteristics. Imprinting, X inactivation; Gene expression regulation and significance in medicine. Prader-willi syndrome, angelman syndrome, * Risk assessment and genetic counseling. Family history in risk assessment. Genetic counseling in clinical practice. Managing the risk for recurrence in families * The treatment of genetic disease. Special consideration in treating genetic disease. Treatment by the manipulation of metabolism. Treatment to increase the function of the affected gene or protein. Modulation of gene expression. Gene therapy. * Application of genomics to medicine and personalizes health care. Pharmacogenetics. Variation in pharmacokinetic response. Adverse drug reaction. Genetic epidemiology. Personalized genomic medicine. | |
| **Textbooks and Materials** |

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| * 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; * 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. |