

Università del Sannio
Corso di Laurea Magistrale in Biologia
Anno accademico 2017-2018

MEDICAL GENETICS
Prof. Giuseppe Iacomino
(6CFU)

GENERAL SECTION:

Structural organization and expression of genes, mutations in the sequences of the genes. Genetic engineering applications in medicine. Animal models. Networks and molecular interactions. Study and analysis of networks pathway interactome. High-throughput technologies and application areas. Genetic counseling, types, methods and tools. Gene therapy. Laboratory diagnosis. Indications for prenatal diagnosis. Collection techniques of fetal material. CVS. Amniocentesis. Cordocentesis. Analysis of material taken and results. Cytogenetic, chromosome analysis, molecular analysis. DNA testing. Genetic counseling in prenatal diagnosis. Preimplantation genetic diagnosis.

GENETIC DEFECTS AND CHROMOSOMAL DISORDERS

Chromosomal abnormalities. Abnormalities of number. Abnormalities of structure. Genomic disorders. Array-CGH and cryptic chromosomal rearrangements. Diagnostic approach to intellectual disability. Sex chromosome abnormalities. Diseases of defects of genomic imprinting: Angelman syndrome. Prader-Willi syndrome. Classification of diseases by dynamic mutations: Fragile X syndrome; Myotonic dystrophy; Huntington's disease; Kennedy disease; Friedreich's ataxia; Spinocerebellar ataxias autosomal dominant cerebellar. Neuromuscular diseases on genetic basis. Spinal muscular atrophies. Amyotrophic lateral sclerosis (ALS). Muscular dystrophies hereditary peripheral neuropathies. Genetic defects in embryonic development. Neurofibromatosis. Tuberous sclerosis. Von Hippel-Lindau. Hemoglobinopathies: structure of hemoglobin. Location, structure and expression of the globin genes. Classification of hemoglobinopathies. Hemoglobinopathies qualitative; Sickle cell anemia; Thalassemia. Inborn errors of metabolism: Cystic fibrosis. Disorders of sexual development. DSD female. DSD male. Genetic causes of infertility. Pharmacogenetics and pharmacogenomics. Nutrigenetics and nutrigenomics. Genetic causes of cancer. Personalized medicine.