



MODELLO SCHEDA INSEGNAMENTO

Corso di L/LM/LMCU	A scelta CORSO DI STUDIO DI LM IN BIOLOGIA CURRICULUM "BIOSANITARIO" e "RISORSE ALIMENTARI E NUTRIZIONE"
Denominazione insegnamento:	Genetica Medica
Numero di Crediti:	6 CFU
Semestre:	Secondo Semestre
Docente Titolare:	Giuseppe Iacomino
Dottorandi/assegnisti di ricerca che svolgono attività didattica a supporto del corso:	
Orario di ricevimento:	da concordare
Indirizzo:	sede

MEDICAL GENETICS

TRAINING

The purpose of course is the acquisition of basic concepts in the field of medical genetics involving the diagnosis and management of hereditary disorders. The increasing impact of genetics in healthcare and the development of newer sophisticated diagnostic technologies will be discussed.

PURPOSES

Acquire the basic concepts of medical genetics, including the interpretation of complex biological systems, genetic counseling and bioinformatics applied, the new techniques of genetic diagnostics with high parallelism, genetic engineering in medicine, examples, hereditary patterns, diagnosis and prevention and genetics clinic Mendelian and complex genetic diseases, including chromosomal syndromes, defects of imprinting, embryological genetic diseases, prenatal diagnosis. Innovative therapies for rare genetic diseases.

PREREQUISITI RICHIESTI

In order to understand and know how to apply most of the topics of this course, it would be beneficial to have a good basic knowledge in the field of cell and molecular biology, genetics, and physiology.

FREQUENZA DELLE LEZIONI

Recommended. Genetic counseling simulations and laboratory trainings will be provided.

CONTENUTI DEL CORSO

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. Genetic causes of cancer. Personalized medicine.

Didactic Methods

The course consists of 48 hours of frontal lessons, which take place in classroom using projections; there are also simple tests at the end of lessons to evaluate the level of understanding.

Reference Texts

RECOMMENDED TEXT: -GENETICA UMANA & MEDICA; G. Neri, M. Genuardi, Edra Masson Ed.
III CONSULTATION TEXTS: -GENETICA MEDICA ESSENZIALE : B.Dallapiccola, G.Novelli
Editore: CIC Edizioni Internazionali. -GENETICA MOLECOLARE UMANA, Pasternak, Zanichelli
Editori. -IL GENE; Lewin; Zanichelli Editori.

Evaluation Method

The achievement of the teaching objectives is certified by passing an oral evaluation exam. The oral exam consists of an interview with questions and discussion on the theoretical contents of the teaching program and is designed to ascertain the level of knowledge and understanding acquired by the student as well as to verify the exposure capacity using the appropriate terminology and the autonomous organization of exposure on the same subjects with theoretical content.

CALENDARIO ESAMI

Rinvio al link

PRENOTAZIONE ESAMI

Rinvio al link