

COURSE SYLLABUS

Genetics and Reproduction

2526-2-F0901D049

Aims

The aim of the course is to provide the student with up-to-date knowledge in the field of medical genetics in all its declinations: from the diagnosis of genetically based pathologies, to the predisposition to tumors, to biotechnologies in the reproductive field, to the evolution of the genome and its implications in human pathologies.

Knowledge and understanding - At the end of the course the student will be able to: Know the pathogenic mechanisms underlying different human genetic pathological conditions; know the genetic markers in the field of prevention, population screening, diagnosis and prognosis of genetic diseases, in the pre-conceptional, pre-natal and post-natal periods; understand the principles of risk and predisposition to human genetic diseases, including tumors; know the genes for sex determination, the biology of human gametes and their use for diagnostic-therapeutic purposes in medically assisted procreation; know the basic principles of population genetics; learn key concepts related to the evolution of the genome.

Ability to apply knowledge and understanding - At the end of the course the student must be able to use the knowledge acquired to understand the potential of medical genetics in the diagnostic and research fields, in relation to the evolution of new technologies in the biomedical field.

Autonomy of judgment - Ability to distinguish the specificities of genetic tests in the diagnosis of genetic diseases, also in the oncology and reproductive fields. Discuss clinical cases; read and critically discuss original works.

Communication skills - ability to use language and terminology in the genetic field.

Learning skills - Ability to correctly interpret the information present in diagnostic reports in the genetic field and in the scientific literature related to medical genetics.

Contents

The course will increase knowledge of Medical Genetics, Genetics of Reproduction; Physiopathology of Reproduction, Gametes and embryology; on associated techniques and technologies in the field of genetics and reproduction.

Detailed program

Medical Genetics: Atypical mechanisms of inheritance: irregular segregation of Mendelian conditions; non-Mendelian segregation of genetic diseases (mitochondrial diseases and genomic imprinting). Medical cytogenetics: Errors of mitotic and meiotic segregation and UPD. Chromosomal syndromes and genomic disorders. Mutations cause of diseases in organization and chromatin remodeling. Diseases caused by dynamic mutations. Principles of multifactorial inheritance. Genetics in oncology practice: hereditary tumor syndromes. genetic testing. Prenatal and postnatal, diagnostic tests, presymptomatic, screening and predictive genetic testing. Genetics of Reproduction: genetics of sex determination and gonadal differentiation. gonadal dysgenesis: genetic causes, sex chromosomes abnormalities and associated syndromes. genetic causes of male and female infertility and of repeat abortions. Human population genetics, natural selection, and genetic conflicts.

Physiopathology of human reproduction and infertility. Epidemiology, diagnostic procedure and medically assisted procreation techniques.

Gametology in human reproduction: spermatogenesis, oogenesis.

Clinical embryology: the process of fertilization. In vitro culture of embryos up to blastocysts. Intrauterine transfer of embryos. Consolidated techniques and innovative techniques.

Techniques of cryopreservation of gametes and germinal tissue. Fertility preservation and social freezing. Biobank management. Pre-implantation genetic diagnosis

The most recent high impact studies in Assisted Reproduction Techniques.

Prerequisites

Basic knowledge in Human Genetics and cell biology

Teaching form

All lessons will be held in person. There are 24 lessons of 2 hours each.

Roughly, 15 of them are frontal lessons.

Ideally 9 lessons will be in seminar format and will be reserved for guests who will talk about their research.

Textbook and teaching resource

Reviews and articles will be given during the course.

recommended books

G. Neri e M. Genuardi. Genetica Umana e Medica. Ed Elsevier (IV ed).

Strachan and Read: human molecular genetics (IV ed)

Semester

first semester- second year

Assessment method

Oral exam. Through a personal bibliographic search, the student proposes to the teachers a scientific article related to the course and asks for its approval within two weeks from the date of the exam. The oral exam consists in the presentation of the article (within a maximum of 10 minutes) and in questions about the rest of the program

Office hours

by appointment only

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Sustainable Development Goals

GOOD HEALTH AND WELL-BEING | QUALITY EDUCATION | GENDER EQUALITY
