



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## COURSE SYLLABUS

### Genetics and Reproduction

2324-2-F0901D049

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#### Aims

The aims of the course is to provide the student with up to date knowledge on i) the pathogenetic mechanisms underlying different human genetic pathological conditions; ii) genetic markers in the field of prevention, population screening, diagnosis and prognosis of genetic diseases, in the preconceptional, prenatal and postnatal period; iii) risk and predisposition to human genetic diseases; iv) genes for the determination of sex; v) the physiopathology of human reproduction; vi) human gametes and their use for diagnostic-therapeutic purposes in medically assisted procreation; vi) genetic predisposition to cancer, with a focus on leukemia predisposition.

#### Contents

The course will increase knowledge of Medical Genetics, Genetics of Reproduction; Physiopathology of Reproduction, Gametes and embryology

#### 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.

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**

Advanced knowledge in Human Genetics and cell biology

## **Teaching form**

classroom lessons

## **Textbook and teaching resource**

Reviews and articles will be given during the course.

G. Neri e M. Genuardi. Genetica Umana e Medica. Ed Elsevier (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 and in questions about the rest of the program

## **Office hours**

by appointment only

## Sustainable Development Goals

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

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