UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Medical Genetics

2122-1-I0302D002-I0302D008M

Aims

The student must be able:

- to recognize the mode of inheritance of Mendelian characters;
- to know the structure of human chromosomes;
- to know the sources of genetic variation;
- to know the mechanisms of epigenetic regulation of gene expression;
- to know basic concepts of quantitative genetics and population genetics.

Contents

The course aims to provide the student with the basis of formal human genetics, introducing the student to the most basic laboratory techniques used for the diagnostic approach and research of hereditary disease.

Detailed program

- Mitosis and meiosis in relation to conventional cytogenetics.
- Mendelian genetics, extensions, recombination and linkage, genetic and physical maps.
- Mendelian Inheritance in man, pedigree reconstruction.
- Sex determination and X chromosome inactivation.

- Fundamentals of epigenetics.
- Polymorphisms and mutations in the context of genetic variability.
- Basic principles of population and quantitative genetics.

Prerequisites

Teaching form

Lectures.

It is required 70% course attendance.

Textbook and teaching resource

Peter J. Russel Genetica Fondamenti

Supplementary material will be provided by teacher

Semester

First semester

Assessment method

Being an integrated course, the evaluation will cover all four modules.

Regarding the Medical Genetics module, the evaluation will consist of a written test that will be used to ascertain

the level of knowledge and ability to understand the topics covered during the course and to be able to solve problems. The student will have to answer 10 quizzes (Multiple choice test) concerning the topics of Medical Genetics.

Oral examination will be required at professor’s discretion (discussion of the written test). The oral test will serve to clarify critical issues emerged from the written test and to verify the communication skills of the student and will focus on the topics covered by the written test.

Office hours

On appointment by e-mail request