UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

# COURSE SYLLABUS

# Human Molecular Genetics

**2021-3-E0201Q073**

## Aims

The course will provide the essential theoretical knowledge in Medical Genetics that will be necessary in future professional applications. It will also allow to obtain a better understanding of the relationships between cell biology, molecular biology, genetics and biomedical sciences.

Knowledge and understanding.
The students shall understand and know: the genetic flow and its regulation; genetic traits inheritance; the relationships between cell biology, molecular biology, genetics and biomedical sciences; molecular mechanisms involved in the generation of phenotypic diversity; molecular mechanisms at the basis of mendelian diseases as well as to complex diseases, in particular those due to epigenetic alterations and those due to interactions with the environment; methods and strategies for the diagnosis of genetic diseases will be illustrated, including new sophisticated techniques.

Ability to apply knowledge and understanding.
The acquired knowledges are functional to understand the basis underlying physiopathological aspects of genetic diseases.

Making judgements.
Students shall be able to connect the different molecular processes at the basis of phenotypic diversity and genetic diseases

Communication skills.
The students are expected to acquire the ability to recognize and describe the above topics, using a proper scientific language;

Learning skills. Learning process will be stimulated by power point presentation highlighting the importance of visual language and images. Learning skills will be also encouraged by discussion of scientific articles on the above topics Examples of examination questions will be proposed to assess the level of comprehension;

## Contents

During the course, basic genetics subjects will be properly analyzed in light of molecular mechanisms, in order to understand the laws of heredity and the processes involved in the generation of phenotypic diversity. Methods and strategies for the diagnosis of genetic diseases will be illustrated, including new sophisticated techniques.

## Detailed program

The course will provide the essential theoretical knowledge in Medical Genetics that will be necessary in future professional applications. It will also allow to obtain a better understanding of the relationships between cell biology, molecular biology, genetics and biomedical sciences. During the course basic genetics subjects will be properly analyzed in order to understand the laws of heredity and the processes involved inthe generation of phenotypic diversity. These concepts will be applied to mendelian diseases as well as to complex diseases, in particular those due to epigenetic alterations and those due to interactions with the environment. Molecular mechanisms responsible for genomic alterations identified by CGH- and SNP-arrays will be described. Main molecular mechanisms that have been already identified and allowed to develop tailored therapy (in oncology or mendelian diseases) will also be illustrated. Among others, the following subjects will be discussed: wild-type, mutated and multiple alleles, dominant and recessive traits; Mendel's laws exceptions: epistasis, penetrance and expressivity; principles and consequences of mitochondrial inheritance and genomic imprinting; multifactorial diseases and quantitative genetics; methods for karyotype analysis; normal karyotype; chromosomal and genomic mutations and their meiotic and phenotypic effect; deletions, inversions, duplications, translocations and non-disjunctions; DNA polymorphisms and their use as genetic markers, in particular related to microarrays; methods and strategies for the diagnosis of genetic diseases.

## Prerequisites

Background: fundamentals of biology and genetics.
Specific prerequisites: Genetics.
General prerequisites: Students can take the exams of the third year after having passed all the exams of the first year of the course.

## Teaching form

During COVID19 emergency periods lecture will be delivered online, with both recorded session and streaming events.

Teaching language: italian.

## Textbook and teaching resource

Learning material (slides of the lessons, recorded lectures, scientific papers, book chapters) is available at the e-learning web page of the course.

Recommended textbook:
Strachan e Read., Genetica Molecolare Umana, Zanichelli 2012

## Semester

First semester

## Assessment method

Written examination (2 h).
Student are asked to answer 20-25 multiple-choice questions and 2 open questions on the whole course content.

During COVID19 emergency period examination will be oral.

## Office hours

Contact: on demand, upon request by mail to lecturer.