



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Genetica Molecolare

2425-1-F0802Q038

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#### Aims

The course aims to provide students advanced knowledge of molecular genetics related to the molecular mechanisms underlying the maintenance of genome stability with particular reference to the identification of potential therapeutic targets and/or diagnostic tools in the field of human health. In addition, knowledge will be provided on the production of mutant variants and on the study of gene-gene and gene-drug interactions, discussing potential biotechnological applications in the industrial field, drug therapy and diagnostics.

Knowledge and understanding. At the end of the course the student will know the mechanisms that maintain the stability of the genome, will be able to produce mutant variants and study gene-gene and drug-gene interactions for biotechnological applications in the industrial field, drug therapy and diagnostics.

Applying knowledge and understanding. At the end of the course the student will be able to apply the knowledge acquired both in the field of basic and applied research.

Making judgment. The student will be able to elaborate what he has learned to apply it to biological problems in which the molecular genetic methods learned can be used.

Communication skills. At the end of the course the student will be able to express himself appropriately in the description of the topics addressed with scientific language properties.

Learning skills. At the end of the course the student will be able to apply the acquired knowledge to problems different from those presented during the course.

#### Contents

The course will focus on the following topics:

- Molecular mechanisms for maintaining genetic stability: DNA repair, homologous recombination, DNA damage tolerance and DNA damage checkpoints. Genetic diseases resulting from their malfunctions. Identification of molecular targets and diagnostic tools for human health.
- Molecular mechanisms regulating telomere stability and consequences derived from their malfunction.
- Genetics screens to identify mutant variants and genetic interactions (positive, negative, gene dosage) to

construct interaction networks for basic and applied research. High-throughput screens for gene-gene and gene-drug interactions to identify new drugs and genetic profiles for personalized drug therapy.

## Detailed program

1. DNA damage and mechanisms of onset of mutations.
2. Mechanisms for DNA damage repair (photoreactivation, BER, NER, MMR, homologous recombination and NHEJ) and genetic diseases associated with their malfunction (eg HNPCC, XP, CS, TTD, Bloom and Werner syndromes). Identifications of potential molecular targets and/or of diagnostic tools for human health with particular interest to anticancer therapies.
3. Mechanisms of DNA damage tolerance: translesion DNA synthesis and homologous recombination.
4. DNA damage checkpoints and genetic diseases resulting from their malfunction (eg AT, ATLD).
5. Genetic controls of telomere stability and consequences of their alterations. Telomerase and proteins of the shelterin complex as possible molecular targets in anti-tumor therapies.
6. Genetic screens after spontaneous or induced mutagenesis for the identification of mutants. Mutation mapping and gene cloning techniques. Random and site-specific mutagenesis. Examples of application for biotechnological purposes on microbial organisms.
7. Genetic screens to identify positive (extragenic suppressors, high dose suppressors) and negative (synthetic lethality) interactions between genes. Genetic analysis of the functional meaning of these interactions and construction of interaction networks. Examples of application for biotechnological purposes.
8. High-throughput screens to identify gene-gene (GGSL) and gene-drug interactions (GCSL) in order to identify new drugs, synergistic effects between drugs and genetic profiles that cause sensitivity or resistance to the action of a drug. Potential biotechnological applications in the field of diagnostics and drug therapy (eg chemotherapy).

## Prerequisites

Background. Basic knowledge of Genetics, Molecular Biology and Biochemistry.

Prerequisites. None

## Teaching form

32 x 2 hours-lectures composed by:

- a section of delivered didactics (Didattica erogativa, DE) focused on the presentation-illustration of contents by the lecturer; each topic is presented in an experimental form, trying to answer the reasons for the experimentation, the way it was carried out, the results achieved and the significance of the results.
- a section of interactive teaching (Didattica Interattiva, DI) including teaching interventions supplementary to delivered didactic activities.

Didactic activities are conveyed by means of face-to-face lectures.

Teaching language: italian.

## Textbook and teaching resource

Slides available at the e-learning platform of the course.

Lectures in the classroom will be recorded and made available in the e-learning page of the course.

Recommended textbooks:

Siede W., Kow Y.W., Doetsch, "DNA damage recognition", Taylor and Francis

Watson J.D., "Biologia molecolare del gene", Zanichelli,

Lewin B., "Il gene VIII", Zanichelli

## **Semester**

Second semester

## **Assessment method**

Oral examination: open questions aimed at verifying the acquired knowledge.

No intermediate evaluations/partial exams.

## **Office hours**

Contact: on demand by mail to the lecturer.

## **Sustainable Development Goals**

GOOD HEALTH AND WELL-BEING | QUALITY EDUCATION

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