



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Fisiopatologia Cellulare

2425-1-F0601Q085

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#### Aims

1. Knowledge and understanding: The course aims to provide an interpretative framework useful to the identification of molecular targets in the diagnosis and therapy of disease and to a critical understanding of research literature
2. Skills in the application of knowledge: the course aims to apply multidisciplinary knowledge, acquired over the entire graduation course, to an integrated vision of disease mechanisms at the cell level
3. Autonomy of judgement: the course aims to provide instruments for critical understanding of scientific literature
4. Communication skills: teaching communication skills, which will be evaluated during the exam, is beyond the scope of the course.
5. Learning skills: at the level of Master Degree, learning skills are given for granted; they will be evaluated during the examination.

#### Contents

The course will focus on themes of cellular pathophysiology with reference to different organic functions . It will deal with both gene-based abnormalities (channelopathies, sarcomere abnormalities etc.) and acquired ones (cell response to stress). To render the course suitable also for students coming from graduation courses other than Biological Sciences, the address to pathophysiology will be preceded by a succinct review of the physiological mechanisms involved..

#### Detailed program

The program is organized around the function of proteins with specific functions (ion channels, trasporters, motor

proteins etc.). The role of these proteins in different organic systems will be illustrated through examples of disease conditions (in parentheses) mechanistically linked to their abnormality. The “mutation” theme will be dealt with, for all listed topics, according to the following scheme: a) protein structure/function, b) protein role in the cell/tissue/organism function; c) mutation site and effect on protein function; d) cellular and organism (syndrome) mutation phenotypes, e) brief address to therapeutic strategies.

Considering the course duration (21 lessons), the topics list (below) is intentionally redundant, i.e. conceived as an “offer”. Since every topic in the list is an independent module, it will be possible to decide during the course which topics to address, depending on students’ background knowledge and interests.

General aspects of membrane e transepithelial transports  
Review of instrumental general physiology concepts

Cellular response to stress  
Intracellular pH homeostasis  
Cell volume homeostasis (RVI and RVD responses)  
Responses to acute and chronic hypoxia/ischemia (syndrome: myocardial ischemia)

Cellular aging

Na<sup>+</sup> channels mutations  
V-gated Na<sup>+</sup> channels (syndromes: LQT3 and Brugada s.)  
Epithelial Na<sup>+</sup> channels (syndromes: pseudo-hypoaldosteronism and Liddle s.)

K<sup>+</sup> channels mutations  
V-gated K<sup>+</sup> channels (syndromes: LQT1 and LQT2, SQT)  
“Inward rectifier” K<sup>+</sup> channels (syndrome: Bartter’s, Andersen-Tawil.)

Cl<sup>-</sup> channels mutations  
Cl<sup>-</sup> channels (syndromes: cystic fibrosis)

H<sub>2</sub>O channels mutations  
Aquaporins (syndrome: diabetes insipidus)

Mutations of proteins involved in intracellular Ca<sup>2+</sup> homeostasis  
RyR channels (syndrome: CPVT)  
SERCA/PLN (syndromes: dilative cardiomyopathy and bronchial asthma)

Ca<sup>2+</sup>-sensing proteins mutations  
Calmodulin (syndromes: LQTS and CPVT)

Motor proteins mutations  
Sarcomeric proteins (syndromes: hypertrophic cardiomyopathy )

## Prerequisites

Acquaintance with the contents of the courses of General and Systems Physiology (graduation course in Biological Sciences) is highly recommended. Depending on students’ background, the teacher will decide the extent to which physiology principles shall be summarized before addressing pathophysiology; this will obviously impact on the number of topics that will be covered in the time available. Considering the impossibility to provide a single reference textbook for the course content, attendance is recommended.

## **Teaching form**

Lectures (delivered component) during which the speaker constantly seeks interaction with the audience (interactive component) through encouragement to hypothesize, to find interdisciplinary connections and, more broadly, to discuss the matter presented by the speaker. The interactive component relies on audience' active participation, to predefine its amount (proportion) is therefore impossible. The implementation of the interactive component requires classroom attendance for the entire course duration; therefore, the remote teaching modality is not considered.

## **Textbook and teaching resource**

A cell Physiology textbook (e.g. Cell Physiology Sourcebook, N. Sperelakis ed., 4th edition, Academic Press) may serve as a reference for basic concepts, but it will not cover many of the specific topics presented in the course. Specific reading material, mostly in the form of review articles, will be made available during the course. Course slides will also be uploaded on the e-learning platform. Unavailability of a comprehensive textbook is a further reason to recommend attendance

## **Semester**

second semester

## **Assessment method**

Students will be evaluated by an oral exam. The exam will focus on knowledge of basic mechanisms; student's ability to apply it to the interpretation of specific conditions and fluency in scientific language will be evaluated. Proactive participation to the interactive component of the course will contribute to a positive judgement. No formal interim evaluations are planned.

## **Office hours**

Please send email to [antonio.zaza@unimib.it](mailto:antonio.zaza@unimib.it) for appointment

## **Sustainable Development Goals**

GOOD HEALTH AND WELL-BEING

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