



UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Cellular Pathophysiology

2324-1-F0601Q085

Aims

The course aims to provide an interpretative framework useful to the identification of molecular targets in the diagnosis and therapy of disease. This course is ideally complementary to those on “Diseases of metabolism ” (biochemical approach) and “Genetic mechanisms of human disease” (genetic approach) in learning about disease mechanisms and therapeutic approaches.

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 parenthesis) mechanistically linked to their abnormality. Considering the course duration (21 lessons), the program below is rather ambitious. However, every topic in the list is an independent module; therefore, it will be possible to decide during the course how many and which topics to address, depending on the students' background knowledge and interest.

Na⁺ channels and their function (review)

Mutations of V-gated Na⁺ channels (syndromes: LQT3 and Brugada s.)
Mutations of epithelial Na⁺ channels (syndromes: pseudo-hypoaldosteronism and Liddle s.)

K⁺ channels and their function (review)
Mutations of V-gated K⁺ channels (syndromes: LQT1 and LQT2, SQT)
Mutations of “inward rectifier” K⁺ channels (syndrome: Bartter s.)

Cl⁻ channels and their function (review)
Mutations of Cl⁻ channels (syndromes: cystic fibrosis)

H₂O channels and their function (review)
Mutations of AQP channels (syndrome: diabetes insipidus)

Intracellular Ca²⁺ homeostasis (review)
Mutations of RyR channels (syndrome: CPVT)
Mutations of SERCA/PLN (syndromes: dilative cardiomyopathy and bronchial asthma)

Ca²⁺-sensor proteins (review)
Mutations of calmodulin (syndromes: LQTS and CPVT)

Motor proteins (review)
Mutations of sarcomeric proteins (syndromes: hypertrophic cardiomyopathy)

Cell response to stress (review)
Acute and chronic response to hypoxia – pathophysiological aspects

Prerequisites

Acquaintance with the contents of the courses of General and Systems Physiology (graduation course in Biological Sciences) is 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

Frontal teaching with interactive discussion

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.

Considering the unavailability of a comprehensive textbook and the interactive teaching modality, attendance to the course is highly recommended

Semester

Second

Assessment method

Students will be evaluated by an oral exam. The exam will focus on knowledge of basic mechanisms and on student's ability to apply it to the interpretation of specific conditions

Office hours

Please send email to antonio.zaza@unimib.it for appointment

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING
