UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Metabolic Diseases

2526-1-F0602Q115

Aims

The biochemical bases of different diseases will be discussed in this course. An interpretative framework will be presented, for the identification of molecular targets in the diagnosis and therapy of diseases, with particular emphasis on common features.

1. Knowledge and understanding: by the end of the course students will have acquired knowledge of the biochemical basis of human metabolic diseases;
2. Applying knowledge and understanding:knowledge acquired in the course will enable students to understand the molecular mechanisms underlying many human diseases;
3. Critical thinking: by the end of the course students will be able to understand biochemical processes in human diseases and apply this knowledge in scientific research;
4. Communication skills: at the end of the course students wil have acquired an adequate scientific language and will be able to properly explain the treated topics.
5. Learning skills: at the end of the course students will be able to read the scientific literature and will have the capacity to pursue further personal studies.

Contents

The course will focus on the most relevant metabolic diseases (aminoacid, sugar, lipid metabolism diseases, lysosomal storage diseases); some neurodegenerative diseases will also be discussed (amyotrophic lateral sclerosis, Parkinson disease, Alzheimer disease, folding diseases). A final part will deal with metabolic alterations in cancer, with particular regards to mitochondrial metabolism.

Detailed program

Aminoacidic metabolism disorders: phenylchetonuria, alcaptonuria, homocystinuria, maple syrup disease, albinism

Nitrogen bases metabolism disorders: Lesch-Nyhan disease, ADA-SCID

Glucose metabolism disorders: G6PDH deficiency. Diabetes

Oxydative Stress*: reactive oxygen species (ROS) and endogenous antioxydant systems

Lipid metabolism disorders: obesity and metabolic syndrome

Heme metabolism disorders: porphyrias

Lysosomal storage diseases: sphyngolipidoses; mucopolysaccaridoses; sialidoses; Pompe disease

Neurodegenerative folding diseases: amyotrophic lateral sclerosis; Parkinson disease, Alzheimer disease. Prion diseases. Trinucleotide repeat disorders

Metabolic alterations in cancer

Mitochondrial metabolism and its alterations

Prerequisites

Acquaintance with the contents of the course of General Biochemistry (graduation course in Biological Sciences) is recommended, particularly as regards glycolysis, Krebs cycle, pentose phosphate shunt, fatty acids synthesis and degradation, ketogenesis and urea cycle. Essential knowledge of General Biochemistry will be summarized before addressing each metabolic disease. Considering the impossibility to provide a single reference textbook for the course content, attendance is recommended.

Teaching form

Didactic teaching with powerpoint presentations (5 cfu)
Interactive teaching with case stuides, kahoots and flipped classroom (1 cfu)

Textbook and teaching resource

Suggested texbooks:

• Cao, Dallapiccola, Notarangelo “Malattie genetiche, molecole e geni” Piccin

• Lieberman e Marks “Biochimica medica, un approccio clinico” CEA

• Leuzzi, Bellocco, Barreca “Biochimica della nutrizione” Zanichelli

Scientific papers will be available on the elearning platform, as well as the slides shown during the course

Semester

First semester

Assessment method

Students will be evaluated through both a written and an oral examination. The written exam will consist of 4 open questions with a length limit. The oral exam, consisting of a discussion of the written exam followed by 1 or 2 short questions, aims at assessing the students' ability to critically discuss common features in different topics.

No *in itinere* evaluations are scheduled.

Office hours

upon email request (paola.fusi@unimib.it)

Sustainable Development Goals

GOOD HEALTH AND WELL-BEING | QUALITY EDUCATION