UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

COURSE SYLLABUS

Metabolic Diseases

2122-1-F0601Q061

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. This course is ideally complementary to those on “Cellular pathophysiology” (physiological approach) and “Genetic mechanisms of human diseases” (genetic approach) in learning about disease mechanisms and therapeutic approaches.

1. Knowledge and understanding - students will acquire knowledge about the most interesting pathogenetic molecular mechanisms, underlying the most common genetic disorders.

2. Applying knowledge and understanding - students will be able to apply the knowledge acquired to the complementary courses in “Cellular pathophysiology” (physiological approach) and “Genetic mechanisms of human diseases”.

3. Making judgements - students will be able to apply the knowledge acquired to experimental research in the field of molecular pathology and also to the evaluation of therapeutic approaches.

4. Communication skills - students will acquire a specific scientific language (both in English and in Italian), as well as the ability to orally describe the topics discussed in the course

5. Learning skills - this course will provide students with the ability to read and critically discuss scientific literature on molecular pathology.

Contents

The course will focus on the most relevant metabolic diseases (aminoacid, sugar and lipid metabolism diseases, lysosomal storage diseases); some neurodegenerative diseases will also be discussed (amyotrophic lateral sclerosis, Alzheimer disease, Parkinson disease, folding disorders). A final part of the course 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

Nucleotides metabolism disorders: Lesch-Nyhan disease, ADA-SCID

Glucose metabolism disorders: G6PDH deficiency. Diabetes

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

Lipid metabolism disorders: obesity and the metabolic syndrome

Heme metabolism disorders: porphyrias

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

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

Metabolic alterations in cancer

Mitochondrial metabolism and its alterations in different disorders

Prerequisites

Acquaintance with the contents of the course in 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 strongly recommended.

Teaching form

Front lectures with powerpoint presentations; interactive lessons and discussions. During Covid emergency all lectures will be telematic and synchronous; all lectures will be recorded and loaded onto the learning platform.

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 Moodle platform, as well as the slides shown during the lessons

Semester

First semester

Assessment method

Students will be evaluated through both a written and an oral examination. The written exam will consist of 5 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)