



UNIVERSITÀ  
DEGLI STUDI DI MILANO-BICOCCA

## SYLLABUS DEL CORSO

### Patologie del Metabolismo

2425-1-F0601Q115

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#### 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.

#### 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: phenylketonuria, 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 antioxydant systems

**Lipid metabolism disorders:** obesity and metabolic syndrome

**Heme metabolism disorders:** porphyrias

**Lysosomal storage diseases:** sphingolipidoses; mucopolysaccharidoses; 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 studies, kahoots and flipped classroom (1 cfu)

## Textbook and teaching resource

Suggested textbooks:

- 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

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