UNIVERSITÀ
DEGLI STUDI DI MILANO-BICOCCA

# COURSE SYLLABUS

# Metabolic Diseases

**2223-1-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 disease” (genetic approach) in learning about disease mechanisms and therapeutic approaches.

## 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, 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

**Nucleotides 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; ethanol toxicity

**Heme metabolism disorders:** porphyrias

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

**Neurodegenerative folding diseases**: amyotrophic lateral sclerosis; Parkinson 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

Frontal teaching with powerpoint presentations; interactive discussions

## 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