UCLouvain

wsbim2230

2024

Biochemistry of inborn errors of metabolism

3.00 credits	30.0 h	Q1

Teacher(s)	Dewulf Joseph (coordinator) ;Nassogne Marie-Cécile ;
Language :	French
Place of the course	Bruxelles Woluwe
Main themes	Genetic basis of inborn errors of metabolism are first reviewed: mutations, patterns of inheritance (autosomal recessive, autosomal dominant, X-linked and mitohondrial transmissions). Next chapters adress the major groups of inborn errors of metabolism : amino-acids, carbohydrates, lipids, purine and pyrimidine, porphyria, neurotransmitters. Peroxisomal, lysosomal and mitochondrial disorders are also described as well as congenital defects of glycosylation.. Each group of diseases are described in relation with the biochemical pathway involved: clinical and biological presentations, physiopathological mechanisms, diagnostic approach, treatment, prognosis and genetic abnormalities. In the last part, inborn errors are reviewed by the way of the study of differential diagnosis of several clinical and biochemical presentations.
Learning outcomes	**At the end of this learning unit, the student is able to :** 1 Introduction to the knowledge of diseases induced by inborn errors of metabolism
Evaluation methods	The exam takes place in two parts: 1. Open book written exam 2. Oral presentation in duo lasting 10 minutes on an article proposed by the teacher
Teaching methods	Interactive course with presentation of theoretical concepts, clinical cases and discussion
Content	The first course recalls the basics of genetics, mutations and their consequences and the modes of transmission of genetic characters: dominant, recessive, sex-linked and mitochondrial. In subsequent courses, the major groups of inborn errors of metabolism will be studied by metabolism: amino acids, carbohydrates, lipids, purines and pyrimidines, porphyria, neurotransmitters. Diseases affecting intracellular organelles, mitochondria, peroxisomes and lysosomes are also described, as are congenital glycosylation defects. Each group of diseases is approached by relating the biochemical pathways concerned, with the clinical and biological presentations, the physiopathological mechanisms, the diagnostic approach, the treatment, the prognosis and the genetic abnormalities. In the last two courses, metabolic diseases are reviewed from the study of the differential diagnosis of clinical and laboratory presentations.
Inline resources	The presentations are available on Moodle
Other infos	Backgrounds : basic knowledge in biochemistry and cellular biology (baccalaureat in medecine, pharmacy or dentistery). PowerPoint are available on icampus.
Faculty or entity in charge	SBIM

Programmes containing this learning unit (UE)				
Program title	Acronym	Credits	Prerequisite	Learning outcomes
Master [120] in Biomedicine	SBIM2M	3		🔍
Master [120] in Biochemistry and Molecular and Cell Biology	BBMC2M	3		🔍
Master [60] in Biomedicine	SBIM2M1	3		🔍
Advanced Master in Clinical Biology	BICL2MC	3		🔍