wsbim2230

2019

In view of the health context linked to the spread of the coronavirus, the methods of organisation and evaluation of the learning units could be adapted in different situations; these possible new methods have been - or will be - communicated by the teachers to the students.

3 credits	30.0 h	Q1

Teacher(s)	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.
Aims	1 Introduction to the knowledge of diseases induced by inborn errors of metabolism - - - - *The contribution of this Teaching Unit to the development and command of the skills and learning outcomes of the programme(s) can be accessed at the end of this sheet, in the section entitled "Programmes/courses offering this Teaching Unit".*
Evaluation methods	**Due to the COVID-19 crisis, the information in this section is particularly likely to change.** Exam : oral evaluation preceeded by written preparation with notes.
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	Aims
Advanced Master in Clinical Biology	BICL2MC	3		
Master [60] in Biomedicine	SBIM2M1	3		
Bachelor in Medecine	MD1BA	3		
Master [120] in Biochemistry and Molecular and Cell Biology	BBMC2M	3		
Master [120] in Biomedicine	SBIM2M	3		
Master [180] in Medecine	MD2M	3		