WSBIM2230

2012-2013

Biochimie des erreurs innées du métabolisme

3.0 credits	30.0 h

Teacher(s) :	Nassogne Marie-Cécile ; Vincent Marie-Françoise (coordinator) ;
Language :	Français
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 :	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 :	Exam : oral evaluation preceeded by written preparation with notes.& bsp;
Other infos :	Backgrounds : basic knowledge in biochemistry and cellular biology (baccalaureat in medecine, pharmacy or dentistery). PowerPoint are available on icampus.
Cycle and year of study :	> Advanced master in Clinical Biology > Master [240] in Medecine > Certificat universitaire de compétence en neurologie pédiatrique > Master [120] in Biomedicine > Master [60] in Biomedicine > Bachelor in Medecine > Master [120] in Biochemistry and Molecular and Cell Biology
Faculty or entity in charge:	SBIM