

Faculty of Medicine



BCMM2130 Biochemistry of Metabolic Diseases

[30h] 2 credits

This course is taught in the 1st semester

Teacher(s): Marie-Cécile Nassogne (coord.), Marie-Françoise Vincent
Language: French
Level: Second cycle

Aims

Introduction to the knowledge of diseases induced by inborn errors of metabolism

Main themes

Genetic basis of inborn errors of metabolism are first reviewed: mutations, patterns of inheritance (autosomal recessive, autosomal dominant, X-linked and mitochondrial transmissions). Next chapters address 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 is 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 review by the way of the study of differential diagnosis of several clinical and biochemical presentations.

Content and teaching methods

This course reviews the clinical, biochemical, genetic and therapeutic aspects of the major inborn errors of metabolism.

Other information (prerequisite, evaluation (assessment methods), course materials recommended readings, ...)

Backgrounds : basic knowledge in biochemistry and cellular biology (baccalaureat in medecine, pharmacy or dentistry).

Exam : oral evaluation preceeded by written preparation with notes.

The course will take place each tuesday of the first half-year : from 4 to 6 pm at ICP 75/-1.

PowerPoint are available on icampus.

Other credits in programs

BIOL22/A	Deuxième licence en sciences biologiques (Biologie moléculaire, cellulaire et humaine)	(2 credits)	
FARM31DS/AN	Première année du diplôme d'études spécialisées en sciences pharmaceutiques (analyses biologiques)	(2 credits)	Mandatory
MED12BA	Deuxième année de bachelier en médecine	(2 credits)	
MED13BA	Troisième année de bachelier en médecine	(2 credits)	
NUT22	Deuxième licence en sciences biomédicales (nutrition humaine)	(2 credits)	Mandatory