Inborn Diseases of Metabolism Affecting Brain Development

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1 ECTS

SUMMARY
Monogenic inborn errors have a prevalence of 1:100. Among these, metabolic diseases affecting brain development have a prevalence of 1:1500. These numerous, but rare and often orphan diseases deeply affect the brain development and functions. This course will provide an overview of the main metabolic diseases affecting brain development, from their genetic to their phenotypic (clinical, pathophysiological and biochemical) description. Students will also learn that these rare diseases also provide an excellent opportunity to analyze brain development and functions from an often unrecognized domain in neuroscience: Intermediary metabolism, which regulate all cellular essential pathways.

DATES IN 2018

The course will take place at the CHUV/PMU Lausanne:

- **Wednesday April 18 from 12h15-14h** (*Salle DELOS, PMU Bugnon, floor 8*)
  - Introduction to the course.
  - Metabolism and cerebral function during development.
  - Isolation and contacts between CNS and periphery: Development of blood-brain barrier and choroid plexus.

- **Wednesday April 25 from 12h15-14h** (*Salle DELOS, PMU Bugnon, floor 8*)
  - Hyperammonemia in newborns and children: Consequences for brain development.

- **Wednesday May 2 from 12h15-14h** (*Salle DELOS, PMU Bugnon, floor 8*)
  - Creatine deficiencies.

- **Wednesday May 9 from 12h15-14h** (*Salle DELOS, PMU Bugnon, floor 8*)
  - Serine deficiencies
  - Deficiency in glucose transporter GLUT1
  - Phenylketonuria and BH4 deficiencies

- **Wednesday May 16 from 12h15-14h** (*Salle DELOS, PMU Bugnon, floor 8*)
  - MCAD deficiency (Medium Chain Acyl-CoA Dehydrogenase)
  - Galactosemia
  - Biotinidase deficiency
  - Non-cetotic hyperglycinemia
  - Homocystinuria

LOCATION
For rooms please see respective course dates; useful maps can be found [here](#).

EVALUATION
Based on a written exam (mini-review).
REGISTRATION
Write an e-mail to the indscourses@gmail.com before March 23, 2018 (course title as subject; supervisor in copy)

READING MATERIALS
Course materials are stored on the UNIL e-learning platform Moodle. You can access by doing the following:

- go to "https://moodle2.unil.ch"
- log in with your institutional/university address
- click on "Faculté de Biologie et de Médecine" > "Ecole doctorale / doctoral school" > "Lemanic Neuroscience Doctoral School"

The materials are stored under "Inborn Diseases of Metabolism Affecting Brain Development". Please use the self-enrollment method to access them.