Inborn Diseases of Metabolism Affecting Brain Development
Organizer: Olivier Braissant

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 2019

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

- **Wednesday May 1 from 12h15-14h** (Salle de séminaire du LCC, CHUV-BH 18-143)
  - 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 May 8 from 12h15-14h** (Salle de séminaire du LCC, CHUV-BH 18-143)
  - Hyperammonemia in newborns and children: Consequences for brain development.

- **Wednesday May 15 from 12h15-14h** (Salle SAMOS, PMU Bugnon, floor 8)
  - Creatine deficiencies.

- **Wednesday May 22 from 12h15-14h** (Salle SAMOS, PMU Bugnon, floor 8)
  - Serine deficiencies
  - Deficiency in glucose transporter GLUT1
  - Phenylketonuria and BH4 deficiencies

- **Wednesday May 29 from 12h15-14h** (Salle SAMOS, 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. The LCC room (BH18-143) is located on 18th floor of the CHUV, within the service (Clinical Chemistry) of O. Braissant. To get there, take one of the speed elevators on the CHUV main floor (8th floor) to floor 18th. To find these ORANGE elevators, turn left at the CHUV main reception and head towards the flower shop ;-) ... Once you are on floor 18, turn to the right (south) and follow the south corridor to its end (if lost,
students can call the course organizer: 079 556 72 07).
The SAMOS room is located on floor 8th of the CHUV (same as main entrance), in the PMU part (policlinique médicale universitaire) next to the public cafeteria.

EVALUATION
Based on a written exam (mini-review).

REGISTRATION
Write an e-mail to the lndscourses@gmail.com before April 1, 2019 (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.