

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 2023

- **Wednesday April 26 from 12h15-14h** (Seminar room HO-1, Hop. Orthopedique, CHUV)
 - 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 3 from 12h15-14h** (Seminar room BH 18-143, 18th floor sud, BH CHUV)
 - Hyperammonemia in newborns and children: Consequences for brain development.
- **Wednesday May 10 from 12h15-14h** (Seminar room HO-1, Hop. Orthopedique, CHUV)
 - Creatine deficiencies.
- **Wednesday May 17 from 12h15-14h** (Seminar room HO-1, Hop. Orthopedique, CHUV)
 - Serine deficiencies
 - Deficiency in glucose transporter GLUT1
 - Phenylketonuria and BH4 deficiencies
- **Wednesday May 24 from 12h15-14h** (Seminar room HO-1, Hop. Orthopedique, CHUV)
 - 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 April 1, 2023 (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.