

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 2025 (location TBA)

Wednesday March 19 from 12h15-14h

- 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 March 26 from 12h15-14h

- Hyperammonemia in newborns and children: Consequences for brain development.

Wednesday April 2 from 12h15-14h

- Creatine deficiencies.

Wednesday April 9 from 12h15-14h

- Serine deficiencies
- Deficiency in glucose transporter GLUT1
- Phenylketonuria and BH4 deficiencies

Wednesday April 16 from 12h15-14h

- MCAD deficiency (Medium Chain Acyl-CoA Dehydrogenase)
- Galactosemia
- Biotinidase deficiency
- Non-cetotic hyperglycinemia

- Homocystinuria

EVALUATION

Based on a written exam (mini-review).

REGISTRATION

Write an e-mail to the indscourses@gmail.com before February 20, 2025 (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.