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 2024 (for room location see here)

- **Wednesday April 10 from 12h15-14h**: Salle Delos, PMU/CHUV, niveau 8 (bâtiment principal)
    - 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 17 from 12h15-14h***:* Salle Delos, PMU/CHUV, niveau 8 (bâtiment principal)
    - Hyperammonemia in newborns and children: Consequences for brain development.
- **Wednesday April 24 from 12h15-14h***:* Salle HO 03, Hôpital orthopédique CHUV (Rue Pierre-Decker)
    - Creatine deficiencies.
- **Wednesday May 1 from 12h15-14h***:* Salle Delos, PMU/CHUV, niveau 8 (bâtiment principal)
    - Serine deficiencies
    - Deficiency in glucose transporter GLUT1
    - Phenylketonuria and BH4 deficiencies
- **Wednesday May 8 from 12h15-14h***:* Salle Delos, PMU/CHUV, niveau 8 (bâtiment principal)
    - 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 lndscourses@gmail.com before March 1, 2024 (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.