

## COURSE DETAILS

### Course description

Neurometabolism is a rapidly expanding field. The recent updated nosology of inborn errors of metabolism (Ferreira et al, 2019) includes more than 1100 diseases and almost 80% of them exhibit neurological symptoms. In particular, during the last 5 years, more than 300 new IEM have been described as emerging causes of prominent neurological dysfunctions. These new disorders have changed paradigms transforming the concept and classification of IEM and are enormously contributing to our understanding of mechanisms in neurological diseases. Most of them stand at the frontier between classical IEM and cellular neurobiology and challenge the traditional organelle-based approach. Additionally, metabolism is a major regulator of brain functions and is involved in many biological processes related to non-metabolic neurological diseases. Therefore, there is a great need for both field's metabolism and neurology, to meet and update previous knowledge. In fact, crucial aspects such as clinical signs (neurological manifestations), pathophysiological categories and therapeutic options need to be redefined.

**Participants are expected to present a case report relevant to the theme of the course; cases with diagnostic and/or therapeutic dilemmas are especially welcome.**

### Learning objectives

The aims of this teaching course are:

- To provide an updated overview of the rapidly growing and changing field of neurometabolic diseases with a special focus on therapies.
- To describe the main neurological manifestations of these diseases at any age (from infants to adults and the old age).
- To describe the biological basis of neurometabolic diseases and provide tools to understand the connection between cell neurobiology and biochemistry. Accordingly, to describe biological concepts such as trafficking, quality control systems (autophagy, molecule repair systems...), signalling...
- To introduce elementary neuroscience concepts of brain physiology.
- To discuss the most recent technical innovations in the diagnosis, characterization and treatment of these diseases (advanced neuroimage techniques, brain stimulation, bioinformatic tools, gene therapy...).

### Scientific organising committee

- José Ramón Alonso, University of Salamanca
- Angeles García-Cazorla, Sant Joan de Déu Hospital, Barcelona
- Fanny Mochel, Pitié-Salpêtrière Hospital, Paris

### Target audience and participant profile

The target audience of this course involves metabolic physicians, paediatricians, neurologists and paediatric neurologists as well as laboratory neuroscientists, biochemical geneticists, biochemists and laboratory geneticists. Participants are expected to have prior knowledge about the field, practical experience with diagnosis treatment, and/or basic research is recommended.

### Fees

The course fees of **450€** cover:

- Hotel accommodation for 2 nights including breakfast.
  - Lunch, coffee and dinner during the course.
  - Course material (pdfs of speakers' presentations)
- A local fee of **315€** is granted if accommodation is not needed.

*Participants are responsible for their own travel arrangements to and from the course.*

*Fees are not refundable.*

### Registration process and deadline

The registration form should be completed on-line: [www.rrd-foundation.org](http://www.rrd-foundation.org) and submitted with your curriculum vitae in English.

No payment is required at this stage.

Deadline for registration is **12<sup>th</sup> of March 2020**.

### Selection criteria and review process

Candidates will be selected based on their background, experience and geographical breakdown.

The scientific organising committee will review the applications and select participants.

Selection decisions will be announced within 10 days following the deadline for registration.

### Accreditation

An application will be made for European CME (EACCME).

**Registration:** [WWW.RRD-FOUNDATION.ORG](http://WWW.RRD-FOUNDATION.ORG)

**Contact:** [CKELLQUIST@RRD-FOUNDATION.ORG](mailto:CKELLQUIST@RRD-FOUNDATION.ORG)

## PROGRAMME

### Thursday 14th May

Start of the meeting at 14:00

#### GENERAL CONCEPTS

**The Human Brain Project. What about metabolism?**

*Felipe Barros, Chile*

**New categories, new diseases: an update in recent relevant findings in neurometabolic diseases**

*Clara D.M. van Karnebeek, Amsterdam*

**Cellular neurometabolism: the connection between metabolism and cell biology in the brain**

*Angeles García-Cazorla, Barcelona*

**Neuroscience Flash - Construction of the human brain: from cells to a complex system**

*José Ramón Alonso, Salamanca*

#### Networking and debate

**Metabolic aspects of the developing brain and neurodevelopmental diseases**

*Soledad Alcántara, Barcelona*

**Brain metabolism of aging and mechanisms of neurodegeneration**

*Fanny Mochel, Paris*

**Clinical Flash - Update in treatable disorders. Don't ever miss a treatable disorder!**

*Clara D.M. van Karnebeek, Amsterdam*

**Participation of a Patient Association representative**

### Friday 15th May

**NEUROLOGICAL MANIFESTATIONS OF IEM (both in children and adults)**

**Early severe encephalopathies**

*Isabelle Desguerre, Paris*

**Non-motor syndromes: "The intellectual disability-psychiatric symptoms-epilepsy spectrum"**

*Angeles García-Cazorla, Barcelona*

**Motor syndromes: from isolated to combined-complex signs (movement disorders, ataxia, spastic paraparesis)**

*Roser Pons, Athens*

**Neuromuscular manifestations**

*Enrico Bertini, Rome*

**Clinical cases from the participants**

**REPRESENTATIVE DISEASES OF THE SIMPLIFIED PATHOPHYSIOLOGICAL BASED CLASSIFICATION**

**Neuroscience FLASH - Understanding mechanisms of memory and cognition**

*José Ramón Alonso, Salamanca*

#### Small molecule defects

Overview: *Jean-Marie Saudubray, Paris*

Focus: **Amino acid synthesis/transport defects:** *Tom J de Koning, Groningen*

#### Complex molecule defects

Overview: *Jean-Marie Saudubray, Paris*

Focus: **Disorders of complex lipid synthesis and remodeling:** *Fanny Mochel, Paris*

#### Energy defects

Overview: *Jean-Marie Saudubray, Paris*

Focus: **GLUT-1 transporter deficiency syndrome:** *Darryl C. De Vivo, New York*

#### Networking and debate

#### WORKSHOPS

- **Neuroimaging**

*Nicole Wolf, Amsterdam*

- **Metabolic Frontiers**

*Saskia Wortmann, Munich*

- **New challenges in treating neurometabolic diseases**

*Fanny Mochel, Paris; Angeles Garcia-Cazorla & Alejandra Darling, Barcelona*

### Saturday 16th May

**THERAPIES IN NEUROMETABOLIC DISEASES. WHAT IS NEW?**

**Neuroscience FLASH - Neurogenesis and neuronal repair**

*José Ramón Alonso, Salamanca*

**Crossing the blood-brain barrier with gate2brain technology**

*Meritxell Teixidó, Barcelona*

**New pharmacological therapies (focused on small molecules)**

*Raphael Schiffmann, Dallas*

**h-Cell therapy/HSCT**

*Florian Eichler, Boston*

**Gene Therapy**

*Alessandro Fraldi, Naples*

**Clinical cases from the participants**

**Brain stimulation. The future of neuromodulation**

*Alejandra Darling, Barcelona*

End of the meeting around 13:00

