

COURSE DETAILS

Course description

Inherited metabolic diseases (IMDs) are an increasing and one of the most common groups among rare diseases. In order to provide optimal care for patients, it is essential that health professionals involved in the diagnosis and management of IMDs have a thorough understanding of these conditions. The purpose of this course is to ensure an appropriate level of knowledge for those training in this area of medicine.

The broad spectrum of IMDs affecting the metabolism of carbohydrates and glycoproteins, amino and organic acids, neurotransmitters, mitochondrial, lysosomal and peroxisomal disorders, will be covered by local and international experts. The programme will include plenary lectures of international experts, case presentations of participants and smaller groups for problem-based learning style, clinical and laboratory workshops.

The course will be interactive and participants are asked to submit a short case study to illustrate a particular diagnostic or clinical management problem.

Learning objectives

- To mediate a basic understanding of human biochemistry and the adverse effects caused by genetic disorders affecting key metabolic pathways.
- To be aware of the variable clinical presentations that occur in IMDs focusing on differences between clinical and management aspects in children, adolescents and adults.
- To appreciate the different analytical techniques used in the diagnosis of IMDs and to know which investigations should be used in order to reach a correct diagnosis.
- To know which disorders are amenable to treatment and consequently require immediate consideration.
- To know the major principles and practicalities of the emergency management of IMDs.
- To appreciate and understand the long-term management of IMDs focusing also on aspects of transitional care and metabolic management during pregnancy.

Scientific Organising Committee

**Centre for Childhood and Adolescent Medicine,
University Hospital Heidelberg**

- Prof. Dr. med. Stefan Kölker
- Prof. Dr. med, Prof. h.c. mult. (RCH) Georg F. Hoffmann
- Priv.-Doz. Dr. med. Dorothea Haas
- Priv.-Doz. Dr. med. Thomas Opladen
- Priv.-Doz. Dr. phil. Jürgen G. Okun

Target audience and participant profile

The course is aimed at paediatricians, physicians, and laboratory scientists aiming to increase their knowledge in the area of IMDs, but is also suitable for specialist dietitians and nurses.

Fees

The course fee of **600€** covers:

- 4 nights hotel accommodation including breakfast
- Lunch, coffee and 2 dinners during the course
- Course material and 2 books

A local fee of **420€** 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 with

1. a curriculum vitae in English,
2. a letter of recommendation from the head of your department,
3. an abstract illustrating a particular diagnostic or clinical management problem.

No payment is required at this stage.

Deadline for registration is **10th of August 2017**.

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 submitted for European CME (EACCME).

PROGRAMME

Tuesday 10th October

| Type of activity | Time | Expert | Topic |
|----------------------------|---------------|----------|---|
| <i>Registration, lunch</i> | 12:00 – 14:00 | | |
| Welcome | 14:00 – 14:15 | Kölker | Introduction to the course |
| Lecture | 14:15 – 15:00 | Zschocke | Quo vadis: the (re-) definition of “inborn errors of metabolism” in the era of massive parallel sequencing |
| Discussion | 15:00 – 15:15 | | |
| Lecture | 15:15 – 16:00 | Haas | When IEM is suspected: Rational diagnostic pathways for “small” and “complex molecule” metabolic diseases |
| Discussion | 16:00 – 16:15 | | |
| <i>Coffee</i> | 16:15 – 16:45 | | |
| Lecture | 16:45 – 17:30 | Gramer | Newborn screening is a programme |
| Discussion | 17:30 – 17:45 | | |
| Lecture | 17:45 – 18:15 | Blau | From symptom to diagnosis assisted by IT |
| Discussion | 18:15 – 18:30 | | |
| Lecture | 18:30 – 19:00 | Hoffmann | Inherited metabolic epilepsies |
| Discussion | 19:00 – 19:15 | | |
| <i>Get together</i> | Start 19:15 | | |

Wednesday 11th October

| Type of activity | Time | Expert(s) | Topic(s) |
|---|---------------|---|---|
| Lecture | 08:30 – 09:00 | Grohmann | Amino acid disorders |
| Presentation | 09:00 – 09:15 | Participant | Case: Amino acid disorder |
| Discussion | 09:15 – 09:30 | | |
| Lecture | 09:30 – 10:00 | Häberle | Urea cycle disorders |
| Presentation | 10:15 – 10:30 | Participant | Case: Urea cycle disorder |
| Discussion | 10:30 – 10:45 | | |
| <i>Coffee</i> | 10:45 – 11:00 | | |
| Lecture | 11:00 – 11:30 | Kölker | Organic acidurias |
| Presentation | 11:30 – 11:45 | Participant | Case: Organic aciduria |
| Discussion | 11:45 – 12:00 | | |
| <i>Lunch</i> | 12:00 – 13:30 | | |
| PBL* groups | 13:30 – 15:00 | Assmann, Opladen Banka, Moog | Meet the experts (2 groups in parallel) W1: Movement disorders W2: Dysmorphology |
| <i>Coffee</i> | 15:00 – 15:30 | | |
| Workshops | 15:30 – 18:00 | Schmidt- Mader, Haas Langhans/ Kohlmüller, Gramer | Lab workshops (2 groups in parallel) W3: Amino acid analysis W4: GC/MS, MS/MS |
| Lab tour | 18:00 – 19:30 | Okun | Lab tour |
| <i>Free evening for participants, speakers dinner</i> | Start 19:30 | | |

*problem-based learning

Thursday 12th October

| Type of activity | Time | Expert(s) | Topic(s) |
|--|---------------|---|---|
| Lecture | 08:30 – 09:00 | Spiekerkoetter | Mitochondrial fatty acid oxidation disorders |
| Presentation | 09:00 – 09:15 | Participant | Case: Fatty oxidation disorder |
| Discussion | 09:15 – 09:30 | | |
| Lecture | 09:30 – 10:15 | Wanders | Non-mitochondrial fatty acid oxidation disorders |
| Presentation | 10:15 – 10:30 | | |
| Discussion | 10:30 – 10:45 | | |
| <i>Coffee</i> | 10:45 – 11:00 | | |
| Lecture | 11:00 – 11:45 | Staufner | Metabolic origin of acute liver failure |
| Discussion | 11:45 – 12:00 | | |
| <i>Lunch</i> | 12:00 – 13:30 | | |
| PBL* groups | 13:30 – 15:00 | Assmann, Opladen Banka, Moog | Meet the experts (2 groups in parallel) W1: Movement disorders W2: Dysmorphology |
| <i>Coffee</i> | 15:00 – 15:30 | | |
| Workshops | 15:30 – 18:00 | Schmidt- Mader, Haas Langhans/ Kohlmüller, Gramer | Lab workshops (2 groups in parallel) W3: Amino acid analysis W4: GC/MS, MS/MS |
| <i>Conference dinner, social programme</i> | 18:00 | | |

Friday 13th October

| Type of activity | Time | Expert(s) | Topic(s) |
|--------------------------------------|---------------|------------------|--|
| Lecture | 08:30 – 09:15 | Burgard | Structured communication to patients and families |
| Discussion | 09:15 – 09:30 | | |
| Lecture | 09:30 – 10:15 | Sahm / Schick | Basic principles of dietary management |
| Discussion | 10:15 – 10:30 | | |
| <i>Coffee</i> | 10:30 – 10:45 | | |
| Lecture | 10:45 – 11:30 | Dionisi-Vici | Basic principles of emergency management |
| Discussion | 11:30 – 11:45 | | |
| <i>Lunch Symposium</i> | 11:45 – 14:00 | All participants | Meeting with patients and their families |
| Lecture | 14:00 – 14:45 | Lachmann | Lost in transition? – When kids reach adulthood |
| Discussion | 14:45 – 15:00 | | |
| <i>Coffee</i> | 15:00 – 15:15 | | |
| Lecture | 15:15 – 16:00 | Murphy | Metabolic management during pregnancy |
| Discussion | 16:00 – 16:15 | | |
| Test | 16:15 – 17:00 | All participants | 20-30 multiple choice questions |
| Lecture | 17:00 – 17:45 | Wijburg | Lysosomal storage disorders |
| Discussion | 17:45 – 18:00 | | |
| <i>Free evening for participants</i> | | | |

Saturday 14th October

| Type of activity | Time | Expert(s) | Topic(s) |
|-----------------------------------|---------------|-----------------|---|
| Lecture | 09:00 – 09:45 | Sperl | Disorders of mitochondrial energy metabolism |
| Discussion | 09:45 – 10:00 | | |
| Lecture | 10:00 – 10:45 | Santer | Disorders of carbohydrate metabolism |
| Discussion | 10:45 – 11:00 | | |
| Test results | 11:00 – 11:45 | Hoffmann/Kölker | Feedback |
| Closing remarks | 11:45 – 12:00 | Hoffmann | Closing remarks and diploma |
| <i>Lunch</i> | 12:00 – 13:00 | | |
| <i>Farewell and Departure</i> | | | |

Registration: WWW.RRD-FOUNDATION.ORG

Contact: CKELLQUIST@RRD-FOUNDATION.ORG