

Prague, 29 February - 2 March 2016

## Course description

Metabolism of sulfur amino acids is intertwined with metabolism and transport of folates and several B-vitamins. This course will give an overview of sulfur amino acids metabolism and of related sulfur containing compounds and its dependence on functional B-vitamin processing. Further topics will include detailed description of inborn errors of metabolism of sulfur amino acids and B-vitamins, association of B-vitamin disturbances with common disorders, diagnostic algorithms for hyperhomocysteinemias and methylation disorders including new-born screening strategies. The organizers and faculty will strongly support discussion about cases presented by the course participants.

## Learning objectives

- Understanding the elements and interactions in the methylation, transsulfuration and remethylation pathways.
- Understanding metabolism of related B-vitamins.
- Knowledge on frequency, pathophysiology, clinical manifestation, diagnosis and treatment of homocystinurias, methylation defects and genetic disorders of B-vitamin metabolism.
- Insight into the role of B-vitamins in common disorders.
- Insight into the principles and efficacy of new-born screening programs, and knowledge on efficacy of early intervention.
- Familiarizing with practical issues in diagnosing and treating patients with CBS deficiency and remethylation disorders.

## Institutes of main organisers - providers

- Charles University in Prague-First Faculty of Medicine and General University Hospital, Institute of Inherited Metabolic Disorders
- Albert-Ludwigs University Freiburg

## Scientific Organising Committee

- Prof. Henk Blom, Albert-Ludwigs- University Freiburg, Freiburg, Germany
- Prof. Brian Fowler, Childrens' University Hospital, Zürich, Switzerland
- Prof. Martina Huemer, Childrens' University Hospital, Zürich, Switzerland
- Prof. Stefan Kölker, University Childrens' Hospital, Heidelberg, Germany
- Prof. Viktor Kožich, Charles University in Prague-First Faculty of Medicine, Prague, Czech Republic

## Target audience and participant profile

The target audience of this advanced course involves metabolic physicians, paediatricians, neurologists, haematologists and other clinical specialist as well as laboratory scientists- biochemical geneticists, biochemists and laboratory geneticists. Participants are expected to have some prior knowledge about the field, practical experience with diagnosis and/or treatment is recommended.

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.

## Fees

The course fees of **450€** cover 2 nights hotel accommodation including the meals during the course. 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 15<sup>th</sup> January 2016.

## Selection criteria and review process

Candidates will be selected based on their background and experience.

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

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

## CME accreditation:

An application will be made to the EACCME for CME accreditation.

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

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

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## PROGRAMME

February 29 is the Rare Disease Day in 2016

### 29<sup>th</sup> of February

Course starts at 14:00

Welcome and introduction

Henk Blom (Freiburg, Germany) and Viktor Kožich  
(Prague, Czech Republic)

#### **Session 1 Introduction and role of E-HOD in improving patient care**

Quiz

Overview of sulfur amino acid and related vitamin  
metabolism

Brian Fowler (Zürich, Switzerland)

E-HOD overview

Henk Blom (Freiburg, Germany)

Patient organization perspective on E-HOD

Marika Groenendijk (Amsterdam, Netherlands)

#### **Session 2 Disorders of methylation and transsulfuration**

Methylation defects-clinical presentation and  
treatment, including outcome E-HOD guidelines on  
methylation defects

Ivo Barić (Zagreb, Croatia)

CBS deficiency-clinical presentation and treatment,  
including outcome E-HOD guidelines on CBS  
deficiency

Andrew Morris (Manchester, UK)

Sulfite oxidase deficiency molybdenum cofactor  
deficiency

Günter Schwarz (Köln, Germany)

### 1<sup>st</sup> of March

#### **Session 3 Remethylation disorders**

MTHFR, cblE and cblG deficiencies

Martina Huemer (Zürich, Switzerland and Bregenz,  
Austria)

CblC and other combined cobalamin defects,  
including outcome E-HOD guidelines on  
remethylation defects

Carlo Dionisi-Vici (Rome, Italy)

#### **Session 4-parallel workshops diagnosing and treating patients with CBS and remethylation defects**

#### **Workshop 1 Diagnosing and treating CBS deficiency**

A.Morris-treatment

V.Kožich-diagnosis

(facilitator - H.Blom)

#### **Workshop 2 Diagnosing and treating remethylation defects**

B.Fowler-diagnosis

M.Huemer-treatment MTHFR,

C.Dionisi-Vici-treatment CblC

(facilitator - M.Baumgartner)

#### **Session 5 Case presentations**

Participants' presentations of "difficult" patients and  
also of undiagnosed patients with abnormal levels of  
methionine, homocystine, B12 or folate.

#### **Session 6 Newborn screening for homocystinurias**

Clinical aspects and E-HOD guidelines on  
homocystinurias and methylation disorders:

Martina Huemer (Zürich, Switzerland and Bregenz,  
Austria)

Technical aspects of screening for homocystinurias:

Viktor Kožich (Prague, Czech Republic)

### 2<sup>nd</sup> of March

#### **Session 7 Homocysteine and B-vitamins in common disorders**

Nutrition and B-vitamins

Anne Molloy (Dublin, Ireland)

Cardiovascular disease and B-vitamin trials

Robert Clarke (Oxford, UK)

B-vitamins and the ageing brain

Helene McNulty (Ulster, Northern Ireland, UK)

Neural tube defects and folates

Henk Blom (Freiburg, Germany)

#### **Session 8 Disorders of cobalamin and folate transport**

Disorders of cobalamin absorption and transport

Ebba Nexø (Aarhus, Denmark)

Disorders of folate absorption and transport

Robert Steinfeld (Göttingen, Germany)

**Final Quiz and discussion – course ends at mid-day**