Metabolic myopathies - Challenges and Solutions

In-person Meeting, Split, Croatia, 16-18 May 2024

Overview

Early diagnosis and management of patients with metabolic myopathies is essential for the patient's outcome. Quick and correct recognition of specific inherited metabolic disease is frequently a challenge both for paediatricians and specialists dealing with adult patients. It may be especially difficult in critically ill patients or patients with multi-organ involvement or neonates and small infants. It requires a multidisciplinary, contemporary and rational approach to distinguish between a primary genetically encoded metabolic disorder, secondary or other muscle disorders resulting in similar clinical picture, particularly in the light of permanently growing number of inherited diseases affecting the muscle and novel diagnostic options.

This interactive course includes lectures, case reports, workshops, debates and patients' stories and is run by a highly specialised and experienced team of experts in metabolic myopathies.

Target Audience

This course is aimed at neurologists, paediatricians and metabolic physicians dealing with muscular diseases, but also geneticists, intensive care specialists, biochemists, physiatrists and other healthcare professionals who are involved in care of patients with muscle diseases.

Scientific Organising Committee

- Ivo Barić, Zagreb, Croatia
- Hanns Lochmüller, Ottawa, Canada
- Danijela Petković Ramadža, Zagreb, Croatia

Learning Objectives

- Understanding essentials of muscle metabolism.
- Understanding pathophysiological, biochemical and genetic bases of metabolic myopathies.
- Getting knowledge on basis, use and potentials of specific tests for diagnosing metabolic myopathies (histology, imaging, electromyography, functional, biochemical and genetic tests).
- Assessment of clinical presentations and clinical situations with possible metabolic myopathy in the background.
- Understanding treatment and management options of patients with metabolic myopathies, including emergency regiments.
- Understanding importance of regular communications with patients and their families.

Venue: Hotel AC Marriott Split

Fees

The fees of 350€ includes :

- 2 nights bed and breakfast,
- Lunches, dinners, guided tour and coffee breaks during the course,
- Speakers' presentations to take away after the course.

Fee of 245€, excluding accommodation, for local participants.

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

Registration Process and Deadline

- The registration form should be completed on www.rrd-foundation.org and submitted with your curriculum vitae and, for those who wish to present a case, an abstract in English.
- No payment is required at this stage.
- Deadline for registration is the 24th of March 2024.

Case presentations by participants

Participants are strongly encouraged to take part in discussions and to submit a case study from their own experience for presentation.

Selection Criteria and Review Process

Candidates will be selected by the scientific organising committee based on background, experience and the geographical breakdown.

CME Accreditation

An application will be made for European CME accreditation.

Click Here to Register

Contact
ckellquist@rrd-foundation.org

PROGRAMME

WITH THE SUPPORT OF : REFERRAL CENTRE FOR GENETICS , METABOLIC DISEASES AND NEWBORN SCREENING OF THE MINISTRY OF HEALTH , REPUBLIC OF CROATIA (DEPARTMENT OF PAEDIATRICS , UNIVERSITY HOSPITAL CENTRE ZAGREB); CROATIAN PAEDIATRIC SOCIETY, SECTION FOR METABOLIC DISEASES

Thursday 16 May

14:30 Start of the course

Introduction and diagnostics of metabolic myopathies

Muscle metabolism
Johannes Mayr, Salzburg

Field of metabolic myopathies
Danijela Petković Ramadža, Zagreb

The role of next-generation sequencing in diagnosing metabolic myopathies
Hanns Lochmüller, Ottawa

Muscle histology/biopsy – important tool despite the era of next generation sequencing
Ichizo Nishino, Tokyo

Neuromuscular imaging in inherited muscular diseases
John Vissing, Copenhagen

Electromyography – a valuable method in diagnosing inherited muscle diseases
Mirea Hančević, Zagreb

Dinner at the hotel

Friday 17 May

Lipid disorders

Fatty acid oxidation and carnitine cycle disorders – focus on the muscles
Ute Spiekerkötter, Freiburg

LPIN and other lipid myopathies
Daniela Tavian, Milan

Case presentations from participants

Friday 17 May continued

Muscle glycogenoses

Muscle glycogenoses: an overview
Benedikt Schoser, Munich

Utility of forearm (non-)ischemic test and exercise testing in diagnostics of metabolic myopathies
John Vissing, Copenhagen

Pompe disease: evolving clinical features and new treatments
Ans van der Ploeg, Rotterdam

Workshops in two groups: Which is optimal diagnostic approach to hyperCKemia?
Moderators: Ros Quinlivan, London; Benedikt Schoser, Munich

Mitochondrial disorders

Overview of mitochondrial myopathies
Saskia Wortmann, Salzburg

Challenges in diagnosing mitochondrial diseases
Johannes Mayr, Salzburg

Workshops in two groups: Mito-cocktails, exercise and nutrition therapy in mitochondrial disease
Moderators: Saskia Wortmann, Salzburg; Rita Horvath, Cambridge

Case presentations from participants

Guided tour and dinner in Split

Saturday 18 May

Some unavoidable topics

Management of patient with severe rhabdomyolysis
Ros Quinlivan, London

Physiotherapy in patients with metabolic myopathies
Jane Newman, Newcastle

Debate: exercise and sports in patients with metabolic myopathies
Moderators: John Vissing, Copenhagen; Jane Newman, Newcastle

Patient stories/testimonials

Other metabolic diseases with predominant muscle involvement

Intersection of metabolic myopathies with other inherited muscle diseases
Rita Horvath, Cambridge

S-adenosylhomocysteine hydrolase deficiency
Ivo Barić, Zagreb

Congenital muscular dystrophies due to glycosylation defects
Ichizo Nishino, Tokyo

13:30 End of the course

Note: Participants are encouraged to prepare case reports of complicated diagnosis or unsolved cases. More information will follow closer to the course.