

INHERITED METABOLIC DISEASE: A CASE BASED APPROACH

IN-PERSON MEETING, HOTEL HOLIDAY INN BLOOMSBURY, LONDON, 22-25 JUNE 2022

OVERVIEW

This course aims to provide an introduction to the **diagnosis and management** of **Inherited Metabolic Diseases** (IMDs). We will use a **case-based approach**, concentrating on the different clinical presentations which can be seen with this diverse group of conditions. For each clinical scenario, we will consider the **differential diagnosis**, **investigations** and **diagnostic pathway**, **acute** and **chronic clinical management**. The format will be **interactive** and will concentrate on working through **clinical examples**, both in **plenary sessions** and smaller group **workshops**, with the help of clinicians, biochemists and dietitians. These are lifelong, genetic diseases and we will cover **paediatric** and **adult** presentations and discuss the lifetime course of disease.

SCIENTIFIC ORGANISING COMMITTEE

- Robin Lachmann, London
- Elaine Murphy, London
- Simon Heales, London
- Helen Prunty, London
- James Davison, London
- Maureen Cleary, London

VENUE

www.hilondonbloomsburyhotel.co.uk

LEARNING OBJECTIVES

- To introduce the different ways in which IMDs can present, both in **children** and **adults**.
- To explore the diagnostic pathway for IMDs with an emphasis on **understanding specialist biochemical investigations**.
- To gain an understanding of the **emergency management** of IMDs.
- To gain an understanding of the **lifetime course of IMDs** and of **disease modifying therapy**.

TARGET AUDIENCE

The course is aimed at clinicians and laboratory scientists who want to **improve their clinical knowledge of IMDs**. As well as those planning a career in IMDs, the course should also appeal to participants from other specialties where IMDs can commonly present (emergency medicine, cardiology, neurology etc.)

FEES

The fees of **500€** includes :

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

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

Fees are only due once candidates have been selected – non refundable.

SELECTION CRITERIA AND REVIEW PROCESS

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

REGISTRATION PROCESS AND DEADLINE

- The registration form should be completed on www.rrd-foundation.org and submitted with your curriculum vitae in English.
- No payment is required at this stage.
- Deadline for registration is the **12th of May 2022**.

COVID-19

Depending on the sanitary situation, local regulations will apply. Stay updated on local regulations [here](#)

CME ACCREDITATION

An application will be made for local CME accreditation.

[CLICK HERE TO REGISTER](#)

CONTACT

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PROGRAMME

Wednesday 22 June

14:00 Start of the course

Session 1: An Introduction to IMD

Archibald Garrod – starting at the beginning, *Robin Lachmann*

Pathogenesis of IMD:

- Metabolites – too much, too little, *Simon Heales*
- Organelle dysfunction, *Shamima Rahman*
- Storage, *Katie Harvey*

Treatment strategies, *Robin Lachmann*

Lifetime disease – how aging affects IMDs, *Simon Heales, Elaine Murphy*

Session 2: Workshops – Laboratory testing (1) (Two of each of these – 45 mins each)

- Amino acids/Organic acids, *Rachel Carling, Helen Prunty*
- Acylcarnitines/VLCFAs, *Amanda Lam, Andrew Morris*
- Enzymology, *Derek Burke, Ralph Wigley, Simon Heales*
- Genetic diagnosis, *Shamima Rahman*

Dinner at the hotel

Note: Participants are encouraged to prepare case reports of complicated diagnosis or unsolved cases. This is not a requirement of registration. More information will follow closer to the course.

Thursday 23 June

Session 2 continued: Workshops – Laboratory testing (2) (Two of each of these – 45 mins each)

- Amino acids/Organic acids, *Rachel Carling, Helen Prunty*
- Acylcarnitines/VLCFAs, *Amanda Lam, Andrew Morris*
- Enzymology, *Derek Burke, Ralph Wigley, Simon Heales*
- Genetic diagnosis, *Shamima Rahman*

Session 3: The first treatable genetic disease

Case presentation – A positive newborn screening result (PKU), *Maureen Cleary*

Introduction to tandem MS, *Helen Prunty*

General NBS talk, *Anupam Chakrapani*

Dietary treatment of aminoacidopathies / tasting session, *Rachel Skeath*

Long-term outcomes and pregnancy, *Robin Lachmann*

New treatment strategies, *Maureen Cleary*

Patient experience, *patient/family*

Session 4: Acute metabolic encephalopathy

Case – Neonatal encephalopathy & differential, *James Davison, Lucinda Carr*

Case – Encephalopathy in adults & differential, *Elaine Murphy, Robin Lachmann*

Session 5: Hypoglycaemia

Case – Ketotic hypoglycaemia & differential, *Andrew Morris, Helen Prunty*

Case – Hypoketotic hypoglycaemia & differential, *Stephanie Grünwald, Helen Prunty*

Dinner in London

Friday 24 June

Session 6: Neurometabolic presentations - workshops (45 mins each)

- Floppy infant, *Jane Hassell, Preeya Rehsi*
- CSF analysis, *Manju Kurian, Simon Heales*
- Muscle presentations, *Mark Roberts*

Session 7: Chronic presentations 1 – workshops (45 mins each)

- Developmental delay, *Apostolos Papandreou, Helen Prunty*
- Hepatosplenomegaly, *James Davison, Anupam Chakrapani, Katie Harvey*
- Dysmorphology, *Stephanie Grünwald, Ali Calder*

Session 8: Chronic presentations 2 – workshops (45 mins each)

- Cardiomyopathy, *Juan Kaski*
- Thrombosis and stroke, *Elaine Murphy, Shamima Rahman, Helen Aitkenhead*
- Psychiatric / behavioural, *Olivier Bonnot*

Dinner at the hotel

Saturday 25 June

Session 9: Hepatic presentations

Case – acute paediatric liver disease, *Roshni Vara, Helen Aitkenhead*

Case – cirrhosis, *Aileen Marshall, Helen Aitkenhead*

Liver transplantation for IMDs, *Aileen Marshall, Roshni Vara*

Session 10: ...and now you are an expert...

Challenging cases, *Led by James Davison (and all faculty)*

13:30 End of the course

