Recordati Rare Diseases Fondation d'entreprise

Inherited disorders of glucose homeostasis
(controversies in hypoglycaemia)
Manchester, UK
14-16 October 2020

COURSE DETAILS

COURSE DESCRIPTION

Hypoglycaemia and hypoglycorrhachia are common in medicine. Failure to recognise a low blood or brain sugar and treat appropriately can lead to severe and permanent damage. This 2 day course for ***metabolic specialists*** *and* ***endocrinologists*** is aimed at exploring some of the more complex, primarily genetic causes, where either diagnosis or management is particularly challenging.
Each topic will take the form of a workshop where brief introductory talks to set the context will be followed by discussion of actual cases where the delegates have found either diagnosis or management to be difficult.
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 review normal mechanisms of glucose homeostasis.
- To review genetic endocrine causes of glucose metabolism, particularly hyperinsulinism and hyperinsulinism-like hypoglycaemias.
- To review inherited metabolic disorders predisposing to hypoglycaemia or hypoglycorrhachia.
- To discuss the aetiology, investigation and management of unexplained hypoglycaemia, in particular 'idiopathic ketotic hypoglycaemia'.
- To agree on a practical approach to investigation and management.

Some of the areas that we plan to cover:

→ *GLUT1 Deficiency Syndrome and other genetic causes of hypoglycorrhachia. This can be a very challenging condition both in terms of diagnosis and management, particularly where patients have been unable to follow a ketogenic diet. How can the diagnosis be confirmed where mutations cannot be identified? What is the experience with the use of alternative treatments such as oral ketones and triheptanoin? How should adult patients be managed.*

→ *Idiopathic ketotic hypoglycaemia. Often this is very straightforward to diagnose and manage but not infrequently there are some very difficult cases where hypoglycaemia and ketosis are severe and frequent. What is the experience of joint endocrine/metabolic clinics for problematic hypoglycaemia?*

→ *Hypoglycaemia in certain fat oxidation disorders. Management of LCHAD – experience with triheptanoin, the need or otherwise for overnight feeding. Experience of rarer disorders such as CACT deficiency.*

→ *Disorders of glycogen storage disorders. In particular GSD1 – how should we manage those children who only tolerate a very short fasting time? What is the role of overnight feeds? How do we prevent hyperinsulinism from excess glucose? What is the role of overnight feeds vs the use of cornstarch? Is it acceptable to run a high lactate and accept slightly lower blood sugars?*

→ *Hyperinsulinism - diazoxide unresponsive patients. Difficulties in diagnosis. Current state of molecular diagnosis.*

→ *Syndromic forms of hypoglycaemia, caused either by mutation in the kinase AKT2 (a critical mediator of insulin action) or activating mutations in PIK3CA or PIK3R2, where the metabolic profile resembles CHI, yet in which neither insulin nor insulin-like molecules can be detected during hypoglycaemia.*

→ *Hypoglycaemia in adults – what are the controversies/difficulties here? Adults with GSD who have developed DM.*

→ *Parents/patients input: How do parents cope with a child GSD1 on 2 hourly feeds? What are the practical difficulties?*

→ *Home glucose monitoring – a blessing or a curse?*

SCIENTIFIC ORGANISING COMMITTEE

- Indi Banerjee, Royal Manchester Children's Hospital
- Beth Jameson, Manchester Centre for Genomic Medicine, St Marys Hospital
- Jean-Marie Saudubray, Consultant in Metabolic Medicine, Pitié-Salpêtrière Hospital, Paris
- John Walter, Manchester Centre for Genomic Medicine, St Marys Hospital

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 and submitted with your curriculum vitae in English.
No payment is required at this stage.

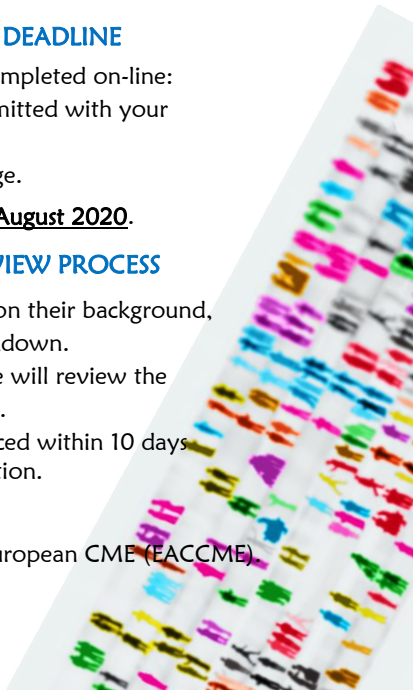
Deadline for registration is 12th of August 2020.

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).



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PROGRAMME

Wednesday 14th October

Start of the meeting at 14:00

Normal mechanisms of glucose homeostasis
Jean-Marie Saudubray

How disorders can affect families
Parent 1 - hyperinsulinism

Parent 2- GSD1

GLUT1 DS
Fanny Mochel, Paris and Emma Glamuzina, Auckland

Hyperinsulinism
Jean-Babtiste Arnoux, Paris and Mark Dunne, Manchester

Thursday 15th October

Disorders of fatty acid oxidation and ketone disorders
Andrew Morris, Manchester

Syndromic hypoglycaemia
Rob Semple, Edinburgh and Sebastian Kummer, Düsseldorf

Disorders of gluconeogenesis and glycogen metabolism
John Walter, Manchester and Philippe Labrune, Paris

Hypoglycaemia in adults
Robin Lachmann, London and Claire Douillard, Lille

Idiopathic ketotic hypoglycaemia, Factitious hypoglycaemia
Beth Jameson and Indi Banerjee, Manchester

Friday 16th October

Participants' cases - Rare causes and difficult cases of hypoglycaemia – adenosine kinase deficiency etc
Participants

A practical (clinical) approach to diagnosis
Jean-Marie Saudubray, Paris

Final discussion and conclusions
John Walter, Manchester

End of the meeting around 12:00

Registration: ***WWW.RRD-FOUNDATION.ORG***

Contact: ***CKELLQUIST@RRD-FOUNDATION.ORG***