Recordati Rare Diseases Fondation d'entreprise

Mitochondrial medicine 30 years on: state of the art
Nice, France
11-13 April 2019

COURSE DETAILS

Course description

Mitochondrial diseases are an important group of inherited metabolic disorders affecting children and adults, with an extremely wide range of clinical symptoms and an ever increasing number of genetic causes.
Since the identification of the first molecular anomalies 30 years ago, the clinical features and molecular basis of the main mitochondrial disorders have been deciphered. Major advances have been achieved in the diagnosis of these disorders in recent years, with the advent of improved molecular techniques.
Despite the lack of curative treatment for most mitochondrial disorders, multidisciplinary care may improve survival and quality of life of patients, and several organ dysfunctions may be treated with specific approaches. In addition, innovative therapies are now available for a few primary mitochondrial diseases, and clinical trials will probably increase in the near future.
An increased awareness of these disorders is therefore needed in order to facilitate earlier diagnosis and genetic counselling for affected families, to improve the multidisciplinary care of patients, and for clinical trial readiness.

Learning objectives

The aims of this teaching course are:

- To describe the pathophysiology, frequency, and clinical symptoms of the main primary mitochondrial disorders presenting in children and adults.
- To describe the main diagnostic tools (exercise tests, muscle biopsy, biochemical and molecular analysis) and to discuss their respective positions in the diagnostic work-up.
- Familiarizing with the differential diagnosis of mitochondrial disease.
- To provide guidance for genetic counselling and reproductive options.
- To describe current treatments and recommended surveillance for patients affected by primary mitochondrial disease.

Scientific Organising Committee

- Prof. Pascal Laforêt, French reference centre for neuromuscular diseases North, East and Paris region - Raymond-Poincaré Hospital, Garches.
- Prof. Shamima Rahman, UCL Great Ormond Street Institute of Child Health, London.
- Dr Angeles Garcia-Cazorla, Neurometabolic Unit/Synaptic Metabolism Lab, Hospital Sant Joan de Déu, Barcelona.

Target audience and participant profile

Pediatricians, neurologists, neuromuscular specialists, internists, cardiologists, geneticists, biochemists, and other clinicians and scientists willing to improve their knowledge of the diagnosis and treatment of primary mitochondrial disorders.

Fees

The course fees of **450€** cover:

- 2 nights hotel accommodation 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 **11th of February 2019**.

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).

Registration: *WWW.RRD-FOUNDATION.ORG*

Contact: *CKELLQUIST@RRD-FOUNDATION.ORG*

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PROGRAMME

Thursday 11 April:

Arrival of delegates

Friday 12 April: Diagnosis

Start of the meeting at 09:00

Session 1: Clinical Recognition of Mitochondrial Disease
Current concepts of mitochondrial disease
Rita Horvath, Newcastle

Overview of mitochondrial disorders in adults
Pascal Laforêt, Paris

Overview of mitochondrial disorders in childhood
Shamima Rahman, London

Session 2: Approaches to Diagnosis of Mitochondrial Disease
Neuro-imaging in mitochondrial disease
Nathalie Boddaert, Paris

Muscle pathology in mitochondrial disease
Edoardo Malfatti, Paris

Genetic diagnosis of mitochondrial disease
Agnes Rotig, Paris

Debate: Muscle biopsy vs Genetics as first line diagnostics for mitochondrial disease
[For] Muscle biopsy first: Enrico Bertini, Rome
[Against] Genetics first: Manuel Schiff, Paris

Session 3: Patient perspective & New diagnostics
Patient perspective

New molecular diagnostics including RNA sequencing
Holger Prokisch, Munich

Proteomics and metabolomics for mitochondrial disorders
To be confirmed

Session 4: Diagnostics workshop
3 parallel workshops (1.5 hours duration):
A) Approach to diagnosis – adult mitochondrial disorders
Pascal Laforêt, Paris + Sabrina Saccone, Nice

B) Approach to diagnosis – paediatric mitochondrial disorders
Shamima Rahman, London + Saskia Wortmann, Salzburg

C) Basic science workshop
Véronique Paquis-Flucklinger, Nice + to be confirmed

Participants to sign up for ONE workshop at time of registration.

Saturday 13 April: Treatment

Session 5: Mitochondrial Therapeutics: Current practice
Participant cases with discussions
Pharmacological approaches to treating mitochondrial disease
Manuel Schiff, Paris

Genetic counselling, PGD and mitochondrial donation for mitochondrial disease
Julie Steffann, Paris

Session 6: Organ involvement in mitochondrial disorders & specific therapies
Epilepsy in mitochondrial disorders: specificities and principles of treatment
Sophie Dupont, Paris

Renal transplant
Stephen Marks, London

Cardiac involvement in mitochondrial disease and heart transplant
Karim Wahbi, Paris

Mitochondrial liver disease and transplantation
Patrick McKiernan, Pittsburgh

The eye in mitochondrial disease and clinical trials
Patrick Yu Wai Man, Cambridge

General discussion about organ involvement in mitochondrial disease

Session 7: Novel therapies & Clinical Trials
Treatment options for MNGIE
Bridget Bax, London

Exercise tests in clinical evaluation and for clinical trials
John Vissing, Copenhagen

Patient registries (including the challenge of global registries)
Michelangelo Mancuso, Pisa

Novel therapies and clinical trials for mitochondrial disease (in adults and children)
Patrick Chinnery, Cambridge

General discussion about novel therapies & clinical trials

End of the meeting around 16:00