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.

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.

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 1st of March 2018.

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).

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.

Contact: CKELLQUIST@RRD-FOUNDATION.ORG
Registration: WWW.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 Proksch, 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