WELLCOME GENOME CAMPUS | ADVANCED COURSES AND SCIENTIFIC CONFERENCES

Mitochondrial Medicine 2018

Wellcome Genome Campus
Hinxton, Cambridge, UK

9-11 May 2018

Draft Conference Programme

Wednesday 9th May 2018

11:30-12:50 **Registration with Lunch**

12:50-13:00 **Welcome and Introductions**
Patrick Chinnery
University of Cambridge, UK

13:00-14:45 **Session 1: Precision diagnostics and new clinical biomarkers**
Chair: Patrick Chinnery, University of Cambridge, UK

13:00 Defining mitochondrial protein function through systems biochemistry
David Pagliarini
Morgridge Institute for Research at UW-Madison

13:30 Probing non-coding variation in Mitochondrial diseases
Holger Prokisch
Institute of Human Genetics, Germany

14:00 Mitochondrial dysfunction of the glutamate pathway revealed by a multi-OMICS approach in MELAS syndrome is alleviated by ketogenic diet
Vincent Procaccio
Angers University, France

14:15 Multivariate metabolic phenotyping for mitochondrial disease diagnosis
Ryan Davis
University of Sydney, Australia

14:30 Background sequence characteristics influence the occurrence and severity of disease-causing mtDNA mutations
Wei Wei
University of Cambridge, UK

14:45-15:00 **Session Discussion**

15:00-15:30 **Afternoon Tea**

15:30-17:00 **Session 2: What determines the phenotype of mitochondrial diseases?**
Chair: *Jan Smeitink, Radboud UMC, The Netherlands*

15:30 Mitochondrial diseases - pathophysiology beyond the organelle
Anu Suomalainen
University of Helsinki, Finland

16:00 Post-transcriptional regulation of mitochondrial metabolism during stress
Elena Rugarli
University of Cologne, Germany

16:30 Post-transcriptional processing of mitochondrial RNA in human disease
Michal Minczuk
MRC Mitochondrial Biology Unit, UK

16:45 In vitro modeling of Leber's hereditary optic neuropathy (LHON) to unravel pathogenic mechanisms and test therapeutic approaches
Valeria Tiranti
IRCCS Institute C. Besta, Italy

17:00-17:15 **Session Discussion**

17:15-18:00 **Lightening Talks** (1 minute per poster – odd number only)

18:00-19:30 **Poster Session I** (odd numbers) **with drinks reception**

19:30 prompt **Dinner**

19:00-23:00 **Cash Bar**

Thursday 10th May 2018

07:30-09:00 **Breakfast**

09:00-10:30 **Session 3: Autophagy-mitophagy: is it important for mitochondrial disease?**
Chair: Massimo Zeviani, MRC Mitochondrial Biology Unit, UK

09:00 Using small molecule modulators for protein import to regulate mitophagy
Carla Koehler
UCLA, USA

09.30 Resolving the pervasive nature of mammalian mitophagy
Ian Ganley
University of Dundee, UK

10:00 Rapamycin rescues a mitochondrial myopathy model via coordinated activation of autophagy and lysosomal biogenesis
Carlo Viscomi
MBU MRC, UK

10:15 Cytochrome C Oxidase deficiency and nuclear DNA damage
Ann Saada (Reisch)
Hadassah-Hebrew University Medical Center, Israel

10:30-10:45 **Session Discussion**

10:45-11.15 **Morning Coffee**

11:15-12:45 **Session 4: Fission/fusion defects in mitochondrial disease: cause or effect?**
Chair: Marni Falk, The Children's Hospital of Philadelphia, USA

11:15 Linking mitochondrial cell biology to mitochondrial diseases
Julian Prudent
MRC Mitochondrial Biology Unit, UK

11:45 Mitochondrial dynamics
Orian Shirihai
Boston University, USA

12:15 Opa1 overexpression ameliorates the renal disease in Mpv17 knockout mice
Marta Luna Sanchez
University of Granada, Spain

12:30 Mitochondrial double stranded RNA triggers antiviral signalling in humans
Ashish Dhir
University of Oxford, UK

12:45-13:00 **Session Discussion**

13:00-14:30 **Lunch**

14.30-16:00 **Session 5: Mitochondrial biogenesis: the key to therapy?**
Chair: Anu Suomalainen, University of Helsinki, Finland

14:30 Strongly impaired mitochondrial CoQ synthesis as a secondary consequence to OXPHOS deficiency caused by reduced mtDNA expression
Nils-Goran Larsson
Karolinska Institutet, Sweden

15:00 NAD(H) sensitive mechanisms in mitochondrial dysfunction
Rong Tian
University of Washington, USA

15:30 Levels of TFAM expression affect mitochondrial function in vivo

Nina Bonekamp
Max Planck Institute for Biology of Ageing, Germany

15:45 GABAergic neurons mediate sudden fatal seizures in a mouse model of Leigh syndrome
Patricia Prada Dacasa
Institut de Neurociències, Autonomous University of Barcelona, Spain

16:00-16:15 **Session Discussion**

16:15-16:45 **Afternoon Tea**

16:45-17:30 **Lightening Talks** (1 minute per poster – even number only)

17:30-18:45 **Poster Session 2** (even numbers) **with drinks reception**

19:00 prompt **Conference Dinner**

19:00-23:00 **Cash Bar**

Friday 11 May 2018

07:30-09:00 Breakfast

09:00-10:30 **Session 6: Gene editing to treat mitochondrial diseases**
Chair: Vasmi Mootha, Harvard Medical School, USA

09:00 Deoxynucleoside therapy for mtDNA depletion disorders
Michio Hirano
Columbia University, USA

09:30 Protein Replacement Therapy for Mitochondrial Genetic Diseases
Haya Louberboum-Galski
Hebrew University, Israel

10:00 MitoRGEN/SpCas9: First Steps Towards Human mtDNA Editing
Natalya Verechshagina
Immanuel Kant Baltic Federal University, Russian Federation

10:15 Genome editing in mitochondria corrects a pathogenic mtDNA mutation in vivo
Payam Gammage
MRC Mitochondrial Biology Unit, UK

10:30-10:45 **Session Discussion**

10:45-11:15 **Morning Coffee**

11:15-12:15 **Session 7: Trial design in rare disease**
Chair: Patrick Chinnery, University of Cambridge, UK

11:15 Clinical trial design for ultra-rare paediatric diseases: challenges for mitochondrial medicine
Shamima Rahman
University College London, UK

11:45 Mitochondrial blindness - light at the end of the tunnel?
Patrick Yu Wai Man
University of Cambridge, UK

12:15-12:45 **Session Discussion**

12:45-13:30 **Session 8: Clinical intervention studies - latest updates**
Chair: Vasmi Mootha, Harvard Medical School, USA

12:45 Treating mitochondrial myopathy with the cyclophilin inhibitor NV556
Magnus Hansson
NeuroVive Pharmaceutical AB, Sweden

13:00 Elamipretide and the MMPOWER Clinical Development Program
Amel Karaa
MGH/Harvard Medical School, USA

13:15 Safety and efficacy of KH176 in adult patients with mitochondrial disease due to the m.3243A>G mutation (KHENERGY): an exploratory, double-blind, randomized, placebo-controlled, two-way cross-over phase II trial
Mirian Janssen
Radboud University MC, Netherlands

13:30 Raxone (idebenone) promotes a clinically relevant recovery of visual acuity in LHON
Catherine Lawrence
Santhera, UK

13:45 **Conference Summary**

14:00 **Conference Closure with Grab Bag lunch**

14:15 Complimentary shuttle to Cambridge train station, Cambridge City Centre, London Stansted Airport and London Heathrow Airport