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**

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**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**

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**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