

Virtual Conference Agenda

Tuesday, 30 November 2021

12:30 12:45 Welcome

[*Marni Falk, The Children's Hospital of Philadelphia, USA*](#)

[*Rita Horvath, University of Cambridge, UK*](#)

[*Mike Patane, Mitobridge Inc. USA*](#)

12:45 13:30 Keynote 1

Introduction to the session

Chair: Marni Falk, The Children's Hospital of Philadelphia, USA

12:45 13:15 Translating Rare Disease Research into Medicine: A 30 year Journey

[*Emil Kakkis, Ultragenyx, USA*](#)

13:15 13:30 Q&A

Chair: Marni Falk, The Children's Hospital of Philadelphia, USA

Moderators: Rita Horvath, University of Cambridge, UK

Mike Patane, Mitobridge Inc., USA

13:30 13:50 Spatial Chat Networking - Meet the speaker & Break

13:50 15:25 Session 1: Mitochondrial Medicine - pharma and funding perspectives

Introduction to the session

Chair: Mike Patane, Mitobridge Inc. USA

13:50 14:05 Academia-pharma interplay in drug development

[*Magnus Hansson, Abliva, Sweden*](#)

14:05 14:20 So, you want to start a mitochondrial company....

[*Amy Ripka, Lucy Therapeutics, USA*](#)

14:20 14:40 Pharma Lightning Talking

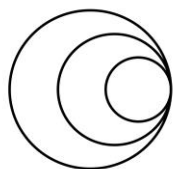
14:40 15:25 Q&A

Chair: Mike Patane, Mitobridge Inc, USA

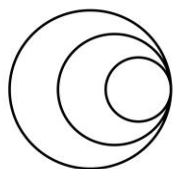
Moderator: Marni Falk, The Childrens Hospital of Philadelphia, USA

Panel Member: Tom Hancock, NMT Venture, USA

15:25 15:55 Spatial Chat Networking - Meet the speakers & Break



15:55	16:55	Poster Session I
15:55	16:55	Poster session I and Spatial Networking
16:55	18:40	Session 2: Pre-clinical mitochondrial disease models 1
		Introduction to the session <i>Chair: Rita Horvath, University of Cambridge, UK</i>
16:55	17:10	Targeting mitochondria: innovation of mitochondrial drug delivery systems (DDS) to mitochondrial medicine <i>Yuma Yamada, Hokkaido University, Japan</i>
17:10	17:25	The fidelity of mitochondrial gene expression <i>Brendan Battersby, University of Helsinki, Finland</i>
17:25	17:40	Generation of novel trans-mitochondrial mice carrying mtDNA with a point mutation in tRNA ^{Leu} (UUR) gene <i>Haruna Tani, University of Tsukuba & Tohoku University, Japan</i>
17:40	17:50	(+)-Epicatechin improves mitochondrial physiology and survival of preclinical C. elegans and zebrafish models of mitochondrial complex I disease <i>Neal Mathew, Children's Hospital of Philadelphia, USA</i>
17:50	18:00	The first in man clinical application of autologous mesoangioblasts as cell therapy medical product in m.3243A>G mutation carriers <i>Florence van Tienen, Maastricht University, Netherlands</i>
18:00	18:40	Q&A <i>Chair: Rita Horvath, University of Cambridge, UK</i> <i>Moderator: Mike Patane, Mitobridge Inc, USA</i> <i>Panel Members: Kaori Ishikawa, University of Tsukuba, Japan</i> <i>Kazuto Nakada, University of Tsukuba, Japan</i>
18:40	19:10	Spatial Chat Networking - Meet the speakers



Wednesday, 1 December 2021

13:00 14:40 Session 3: Clinical trials - validation of outcome measures

Introduction to the session

Chair: Shamima Rahman, UCL, Great Ormond Street Institute of Child Health, UK

13:00 13:10 Clinical trial readiness for Leigh Syndrome: The International Leigh Syndrome Roadmap Project

[*Shamima Rahman, UCL, Great Ormond Street Institute of Child Health, UK*](#)

13:10 13:25 New myopathy outcome measures in the Mitochondrial disease clinical trials era

[*Zarazuela Zolkipli-Cunningham, Children's Hospital of Philadelphia, USA*](#)

13:25 13:40 Clinical trial endpoints for an ultra-rare genetic disorder: Barth syndrome as a model

[*Hilary Vernon, John Hopkins University, USA*](#)

13:40 13:50 An open-label study to evaluate the safety and tolerability of 12 weeks treatment with oral REN001 in patients with primary mitochondrial myopathy (PMM), with an optional extension of treatment

Grainne Gorman, Newcastle University, UK

13:50 14:00 The natural history of BCS1L related mitochondrial disease: a multinational cohort study of 33 cases

Omar Hikmat, Haukeland University Hospital, Norway

14:00 14:40 Q&A

Chair: Shamima Rahman, UCL Great Ormond Street Institute of Child Health, UK

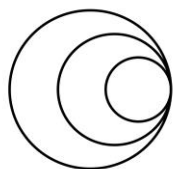
Moderator: Mike Patane, Mitobridge Inc. USA

14:40 15:10 Spatial Chat Networking - Meet the speakers and Break

15:10 16:00 Lightning Talks and Poster Session II

15:10 15:25 Poster session II lightning talks

15:25 16:00 Poster session II and Spatial Networking



16:00 18:00 Session 4: Therapeutic development in mitochondrial eye disease

Introduction to the session

Chair: Jose Sahel, UPMC, USA

16:00 16:15 Gene Therapy for Leber Hereditary Optic Neuropathy

[Patrick Yu Wai Man, University of Cambridge, UK](#)

16:15 16:30 Measuring Mitochondrial Dysfunction in Retinal Dystrophies

[Kurt Rieger, OcuMet, USA](#)

16:30 16:45 Exploiting hiPSCs in Leber's hereditary optic neuropathy (LHON): a challenging opportunity

[Valeria Tiranti, IRCCS Institute C. Besta, Italy](#)

16:45 16:55 Genotype-Phenotype Correlations in Autosomal Dominant Optic Atrophy

Joshua Harvey, Institute of Ophthalmology, UK

16:55 17:05 The Phase III REFLECT Trial: Efficacy and Safety of Bilateral Gene Therapy for Leber Hereditary Optic Neuropathy (LHON)

Mark Moster, GenSight Biologics, France

17:05 17:15 Therapeutic 670nm light protection of retinal ganglion cell degeneration in OPA1 dominant optic atrophy mouse

Marcela Votruba, Cardiff University, UK

17:15 17:55 Q&A

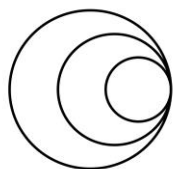
Chair Jose Sahel, UPMC, USA

Moderator: Rita Horvath, University of Cambridge, UK

Panel Member: Lissa Poincenot, LHON, USA

Russell Wheeler, The LHON Society, UK

17:55 18:25 Spatial Chat Networking - Meet the speakers



Thursday, 2 December 2021

13:00 14:45 Session 5: Pre-clinical mitochondrial disease models 2

Introduction to the session

Chair: Marni Falk, The Children's Hospital of Philadelphia, USA

13:00 13:15 Mouse Models of Mitochondrial Disease

[*Douglas Wallace, Children's Hospital of Philadelphia, USA*](#)

13:15 13:30 Engineering the Intracellular Microbiome: Lessons from Zebrafish

[*Stephen Ekker, Mayo Clinic, USA*](#)

13:30 13:45 Exploring mitochondrial function in *C. elegans*: automated high-content phenotyping with the "Worm-on-Chip" technology

[*Elena Katsyuba, Nagi Bioscience, Switzerland*](#)

13:45 13:55 Preferential Propagation of Mitochondrial DNA Mutation During Oocyte Development

Haixin Zhang, University of Cambridge, UK

13:55 14:05 The soluble guanylate cyclase stimulator CY6463 improves neuromuscular function and swimming activity in multiple zebrafish models of mitochondrial respiratory chain disease

Leonard Burg, Children's Hospital of Philadelphia, USA

14:05 14:45 Q&A

Chair: Marni Falk, The Children's Hospital of Philadelphia, USA

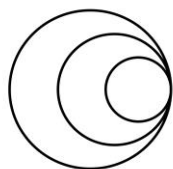
Moderator: Rita Horvath, University of Cambridge, UK

14:45 15:25 Spatial Chat Networking - Meet the speakers and Break

15:25 16:25 Lightning talks and Poster Session III

15:25 15:40 Poster session III lightning talks

15:40 16:25 Poster session III and Spatial Networking



16:25 17:55 Session 6: Collaboration and data sharing to support precision medicine

Introduction to the session

Chair: Xiaowu Gai, Children's Hospital Los Angeles, USA

16:25 16:40 Databases of mitochondrial genes and their variation: MitoCarta3.0 and gnomAD v3

[*Sarah Calvo, Broad Institute of MIT/Harvard, USA*](#)

16:40 16:55 A reference atlas of mitochondrial genotype-phenotype associations in the UK Biobank

[*Claudia Calabrese, University of Cambridge, UK*](#)

16:55 17:05 Targeted Therapies for Leigh Syndrome: Systematic Review and Steps Towards a 'Treatabolome'

May Yung Tiet, Horvath Laboratory, UK

17:05 17:15 Precision mitochondrial medicine in practice: CHOP MMFP-Tableau data warehouse guides Therapeutic development

Ibrahim George-Sankoh, Childrens Hospital of Philadelphia, USA,

17:15 17:55 Q&A

Chair: Xiaowu Gai, Children's Hospital Los Angeles, USA

Moderator: Marni Falk, The Children's Hospital of Philadelphia, USA

17:55 18:25 Spatial Chat Networking - Meet the speakers and Break

18:25 18:35 Closing remarks

Scientific Programme Committee:

[*Marni Falk, The Children's Hospital of Philadelphia, USA*](#)

[*Rita Horvath, University of Cambridge, UK*](#)

[*Mike Patane, Mitobridge Inc. USA*](#)