

Hybrid Conference Programme

Start (GMT)	Finish (GMT)	Presenter details
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Monday 21 November 2022

11:30 12:50 Registration, lunch and networking

12:30 Briefing for Keynote & Session 1 speakers, chair, moderator & mic runners - Auditorium

12:50 13:00 Welcome

Scientific Programme Committee:

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

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

[Michelangelo Mancuso, University of Pisa, Italy](#)

13:00 14:00 Keynote 1

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

Moderator: [Michelangelo Mancuso, University of Pisa, Italy](#)

13:00 14:00 Gene and Mutation-Specific Precision Therapies: From GAN to the COL6 Related Dystrophies and Beyond
[Carsten Bonnemann, NIH, USA](#)

14:00 14:10 Comfort break - (refreshments available at 15:40)

14:10 15:40 Session 1: Bridging Academia and Industry

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

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

14:10 14:40 From preclinical models to biomarkers and clinical trials for mitochondrial disease
[Anu Suomalainen, University of Helsinki, Finland](#) - VIRTUAL

14:40 15:10 Mitochondrial genome engineering in vivo
[Michal Minczuk, University of Cambridge, UK](#)

15:10 15:16 Mitochondrial augmentation technology: Harnessing mitochondria to improve cellular functionality
[Natalie Yivgi Ohana, Minovia Therapeutics, Israel](#)

15:16 15:22 Mitochondrial DNA replaced hematopoietic stem/progenitor cells (LSK cells) treatment to a mouse model of heteroplasmic mtDNA mutation showed increased wild type tRNA-Ala levels in mice tissues
[Dori Pelled, Imel Biotherapeutics Inc, USA](#) - VIRTUAL

15:22 15:28 Results from a Phase 2a study evaluating CY6463, a CNS-penetrant sGC stimulator, in adults with MELAS
[Chad Glasser, Cyclerion Therapeutics, USA](#) - VIRTUAL

15:28 15:34 The soluble guanylate cyclase stimulator CY6463 improves mitochondrial physiology, maximal swimming capacity and whole body oxygen consumption capacity with exercise in a surf1^{-/-} zebrafish model of mitochondrial complex IV deficiency
[Leonard Burg, Children's Hospital of Philadelphia, USA](#) - VIRTUAL

15:34	15:40	Analysis of an Increased Dose of Elamipretide in Patients with Rare Diseases: Safety and Efficacy Results from the Expanded Access Program <i>Mary Kay Koenig, UT Health & Stealth, USA - VIRTUAL</i>
15:40	16:10	Refreshment break
16:00		<i>Briefing for Session 2 speakers, chair, moderator & mic runners - Auditorium</i>
16:10	17:10	Session 2: Putting Patients at the Heart of Clinical Trials - Panel Session
		<i>Chair: Michelangelo Mancuso, University of Pisa, Italy</i> <i>Moderator: Marni Falk, The Children's Hospital of Philadelphia, USA</i>
16:10	16:25	The Top Ten Research Priorities for Rare Mitochondrial Diseases: Results of a Patient/Health Professional Priority Setting Partnership <i>Marcela Votruba, Cardiff University, UK</i>
16:25	16:40	Patterns and barriers to clinical trial participation for patients with primary mitochondrial disease: A survey of patients/caregivers, physicians, and pharmaceutical industry professionals <i>Philip Yeske, UMD, USA- VIRTUAL</i>
16:40	17:10	Panel session
17:10	17:25	Comfort break
17:15		<i>Briefing for Session 3 speakers, chair & moderator - Auditorium</i>
17:25	19:25	Session 3: Clinical Trials Readiness
		<i>Chair: Marni Falk, The Children's Hospital of Philadelphia, USA</i> <i>Moderator: Michelangelo Mancuso, University of Pisa, Italy</i>
17:25	17:55	Leigh syndrome: the experience of Beijing Children's Hospital <i>Fang Fang, National Center for Children's Health, China - VIRTUAL</i>
17:55	18:25	Natural history of mitochondrial optic neuropathies <i>Thomas Klopstock, University of Munich, Germany</i>
18:25	18:55	Prospective natural history study (CLARITY) and longitudinal nutritional assessment in mitochondrial disease <i>Zarazuela Zolkiplo-Cunningham, The Children's Hospital of Philadelphia, USA</i>
18:55	19:25	Panel Discussion
19:25		Dinner
19:25		Bar open (card payments only)

Tuesday 22 November 2022

07:30 09:00 Breakfast

09:25 Briefing for Session 3 speakers, chair & moderator - Auditorium

09:40 10:20 Lightning talks for virtual posters

Chair: Michelangelo Mancuso, University of Pisa, Italy

10:20 11:20 Session 4: Therapeutic Development for Mitochondrial Disease

Chair: Michelangelo Mancuso, University of Pisa, Italy

Moderator: Rita Horvath, University of Cambridge, UK

10:20 10:50 Current State of Corporate Activity in Mitochondrial Medicine Therapeutic Development
[Marni Falk, The Children's Hospital of Philadelphia, USA](#)

10:50 11:00 The selective autophagy of mitochondria
Michael Clagu, University of Liverpool, UK

11:00 11:10 The effects of resistance exercise training on mitochondrial myopathy patients
Valeria Di Leo, Newcastle University, UK

11:10 11:20 Intra- and Extracellular Metabolic Investigation of Perfused Fibroblasts by Real-time NMR Allows for Better Discrimination of Mitochondrial Defects
Christian Urzi, University of Bern, Switzerland

11:20 11:30 Developing a mitochondrial gene therapy for MELAS: Editing the MT-TL1 locus in human cell lines harboring the m.3243A>G pathogenic variant
Santiago Castillo, Mayo Clinic, USA - VIRTUAL

11:20 12:00 Lightning talks for in-person posters

Chair: Michelangelo Mancuso, University of Pisa, Italy

12:05 14:05 Lunch with posters

13:55 Briefing for Session 4 speakers, chair, moderator & mic runners - Auditorium

14:05 15:35 Session 5: Multiomics Data Analysis

Chair: Rita Horvath, University of Cambridge, UK

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

14:05 14:35 Integrative analysis of multi-omics data to advance diagnostics
Holger Prokisch, Institute of Human Genetic of the Technical University Munich, Germany

14:35 15:05 A diagnosis for all rare genetic diseases: The horizon and next frontiers
[Kym Boycott, University of Ottawa, Canada](#)

15:05 15:15 Defining the nuclear genetic architecture of a maternally-inherited mitochondrial disorder
Roisin Boggan, Wellcome Centre for Mitochondrial Research, UK

15:15 15:25 National mitochondrial disease registry in England: linking genetics with routinely collected healthcare data
Katherine Schon, University of Cambridge, UK

15:25 15:35 Expanding MitoPhen: an overview of preliminary results
Thiloka Ratnaile, Colchester Hospital, UK - VIRTUAL

15:35 16:20 Refreshment break

16:10 Briefing for Session 6 speakers, chair, Reneo representative, moderator & mic runners - Auditorium

16:20 17:50 Session 6: Exploring Brain Phenotypes and Treatments

Chair: Rita Horvath, University of Cambridge, UK

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

- 16:20 16:50 The randomized controlled phase 2b KHENERGYZE trial of sonlicromanol in primary mitochondrial m.3243A>G disease patients: topline results
[Jan Smeitink, Khondrion, Netherlands](#)
- 16:50 17:20 Brain and Mitochondria: The role of energy metabolism on mood disorders
[Ana Andreazza, University of Toronto, Canada](#)
- 17:20 17:30 Developing mouse models to fight POLG-related diseases
Carlo Viscomi, University of Padova, Italy
- 17:30 17:40 Lutein rescues a nlg-1-mediated synaptic defect in a C. elegans mitochondrial Complex I deficiency model
Silvia Maglioni, IUF-Leibniz Research Institute for Environmental Medicine, Germany
- 17:40 17:50 N-acetylcysteine and cysteamine bitartrate antioxidant therapies prevent neuromuscular decompensation in two novel surf1^{-/-} zebrafish models of Leigh Syndrome
Suraiya Haroon, The Children's Hospital of Philadelphia, USA
- 17:50 18:50 Sponsored Presentation and Drinks Reception - Reneo Pharma Ltd, UK
- 18:50 Dinner
- 18:00 Bar open (card payments only)

Wednesday 23 November 2022

07:30 09:00 Breakfast

09:15 Briefing for Session 7 speakers, chair, moderator & mic runner - Auditorium

09:30 11:00 Session 7: The Role of Mitochondria in Cancer

Chair: Michelangelo Mancuso, University of Pisa, Italy

Moderator: Rita Horvath, University of Cambridge, UK

09:30 10:00 Mitochondrial DNA mutations in ageing and cancer - What's the connection?

[Laura Greaves, Newcastle University Centre for Cancer, UK](#)

10:00 10:30 Reduction of OXPHOS capacity with small molecule inhibitors of mtDNA transcription to treat metabolic

[Nils-Garon Larsson, Karolinska Institutet, Sweden](#)

10:30 10:45 Impaired energy metabolism in Fanconi Anemia: role of imbalanced mitochondrial dynamics

Nadia Bertola, University of Genoa, Italy - VIRTUAL

10:45 11:00 *Role of ClpP and mitochondrial metabolism in the anti-cancer effects of imipridone ONC201 and ONC206 in glioblastoma and DIPG*

Varun Prabhu, Chimerix, USA - VIRTUAL

11:00 11:45 Refreshment break

11:30 Briefing for Ssession 8: Scientific Programme Committee, panel members & mic runners - Auditorium

11:45 12:30 Session 8: Challenges and opportunities in mitochondrial medicine - perspectives from pharma and funders

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

Moderator: Michelangelo Mancuso, University of Pisa, Italy

Join panelists for a live discussion on current challenges and future opportunities in mitochondrial medicine research and therapies

12:30 12:45 Closing remarks and prize presentation

Scientific Programme Committee:

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

Michelangelo Mancuso, University of Pisa, Italy

12:45 13:30 Lunch and departures

13:30 Coach departures for Stansted and Heathrow airports

13:40 Coach departures for Cambridge train station and city centre