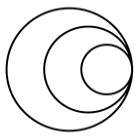


Hybrid Conference Programme

Start	Finish	Presenter details
Monday 18 March 2024		
12:00	12:55	Registration, lunch and networking
12:40		Briefing for Keynote & Session 1 speakers, microphone runners, chair, moderator & committee - Auditorium
12:55	13:10	Welcome
		<p>Wellcome Connecting Science: Nagehan Ramazanoglu Bahadir, Wellcome Connecting Science, UK</p> <p>Scientific Programme Committee: Rita Horvath, University of Cambridge, UK Amel Karaa, Massachusetts General Hospital/Harvard University, USA Nils-Göran Larsson, Karolinska Institute, Sweden Michelangelo Mancuso, University of Pisa, Italy</p>
13:10	14:10	Keynote I
		<p>Chair: Rita Horvath, University of Cambridge, UK Moderator: Cristina Dominguez-Gonzalez, Spain</p> <p>Mitochondrial medicine: lessons learned from population genomics Patrick Chinnery, University of Cambridge, UK</p>
14:10	14:15	Comfort break
14:15	15:45	Session 1: Basic science to treatment: MtDNA replication defects
		<p>Chair: Nils-Göran Larsson, Karolinska Institute, Sweden Moderator: Nicole Lake, Yale University, USA</p>
14:15	14:45	<p>Nucleotide metabolism as a therapeutic target for mtDNA maintenance disorders Ramon Marti, Vall d'Hebron Institut de Recerca, Spain</p>
14:45	15:15	<p>Molecular consequences of POLG mutations and new avenues of treatment Maria Falkenberg, University of Gothenburg, Sweden</p>
15:15	15:30	<p>Deoxyguanosine kinase deficiency: natural history and liver transplant outcome Caterina Garone, University of Bologna, Italy</p>
15:30	15:45	<p>Metformin Ameliorates Pathogenic Phenotypes in POLG-Related Encephalopathy Organoids: A Novel Mitochondrial Medicine Approach Kristina Xiao Liang, University of Bergen, Norway</p>
15:45	16:30	Refreshment break and networking
16:15		Briefing for Session 2 speakers, chair & moderator - Auditorium



16:30 18:00 Session 2: Basic science to treatment II: MtDNA transcription and translation

Chair: Rita Horvath, University of Cambridge, UK

Moderator: Caterina Garone, University of Bologna, Italy

16:30 17:00 Modulating metabolism to treat mitochondrial dysfunction

[*Aleksandra Filipovska, University of Western Australia, Australia*](#)

17:00 17:30 Nutrition in a basket: Amino-acids to treat ARS1 deficiencies

[*Clara van Karnebeek, Amsterdam UMC, Netherlands*](#)

17:30 17:45 Inhibition of mtDNA transcription reverses diet-induced obesity and drives systemic metabolic benefits in mice

Taolin Yuan, Karolinska Institute, Sweden

17:45 18:00 Estrogens rescue mitochondrial dysfunction in iPSCs induced by m.3243A>G mutation

Danyang Li, Oxford University, UK

18:00 18:40 Poster pitch talks for odd number posters

18:40 19:40 Poster session 1 - odd number posters with drinks reception

19:40 Dinner

Bar open (card payments only)

Tuesday 19 March 2024

09:15 Briefing for Session 3 speakers, chair & moderator - Auditorium

09:30 11:00 Session 3: Basic science to treatment III: primary MtDNA defect

Chair: Michelangelo Mancuso, University of Pisa, Italy

Moderator: Patrick Yu-Wai-Man, University of Cambridge, UK

09:30 10:00 An in vivo genetic screen identifies cellular pathways to rescue the deleterious effects of mitochondrial DNA mutations

[Anna Wredenberg, Karolinska Institute, Sweden](#)

10:00 10:30 Fixing mtDNA disorders: The future of clinical trials

[Amel Karaa, Massachusetts General Hospital/Harvard University, USA](#)

10:30 10:45 Effects of the novel therapeutic SBT-589 across models of Friedreich's ataxia

Laura Kropp, Stealth Bio Therapeutics, USA

10:45 11:00 Primary mitochondrial disease causing mtDNA variants in the context of African (L) haplogroups

Surita Meldau, University of Cape Town/National Health Laboratory Services, South Africa

11:00 11:45 Refreshment break and networking

11:30 Briefing for Session 4 speakers, chair & moderator - Auditorium

11:45 13:15 Session 4: Gene editing and therapies

Chair: Kathrin Meyer, Nationwide Childrens Hospital, USA

Moderator: Aleksandra Filipovska, University of Western Australia, Australia

11:45 12:15 The Use of Mitochondrial-Targeted Gene Editing Enzymes as Therapy for Mitochondrial Diseases

[Carlos Moraes, University of Miami Health System, USA](#)

12:15 12:45 Mitochondrial Transplantation Therapy, Recent Advances and Perspective

[James McCully, Boston Children's Hospital/Harvard University, USA](#)

12:45 13:00 AAV-based liver-targeted gene therapy for MNGIE: proposal for a clinical trial

Jelle van den Ameele, University of Cambridge, UK

13:00 13:15 A gene edited mouse model with a pathogenic mtDNA mutation in tRNA-Glu causing kidney disease

Min Jiang, Westlake University, China

13:15 14:00 Lunch

14:00 14:30 Dessert and refreshments within the exhibition space

14:15 Briefing for Session 5 speakers, chair, moderator - Auditorium

14:30 15:50 Session 5: Panel discussion - How do we rally the international community for clinical trial consortia?

Chair: Michelangelo Mancuso, University of Pisa, Italy

Moderator: Amel Karaa, Massachusetts General Hospital/Harvard University, USA

14:30 14:50 PRECISION ALS : A Bespoke Patient Data Platform and Analytic System for Amyotrophic Lateral Sclerosis

[Orla Hardiman, Trinity College Dublin/ENCALS, Ireland - VIRTUAL](#)

14:50 15:10 Drug regulatory processes in the UK and the future of Rare Disease Drug Development

[Viji Ajith, Medicines and Healthcare Products Regulatory Agency, UK - VIRTUAL](#)

15:10 15:20 *Introductory presentations*

Alison Maguire, The Lily Foundation, UK

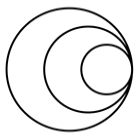
Philip Yeske, United Mitochondrial Disease Foundation, USA

15:20 15:50 *Panel discussion to also include:*

Sean Murray, Mito Foundation, Australia

Brian Tseng, The PoIG Foundation, USA

15:50 16:35 Refreshment break and networking



16:20 Briefing for Session 6 speakers, chair, moderator - Auditorium

16:35 17:20 Session 6: Panel discussion - Challenges and opportunities in clinical trials: perspectives from pharma and funders

Chair: Amel Karaa, Massachusetts General Hospital/Harvard University, USA

Moderator: Michelangelo Mancuso, University of Pisa, Italy

Confirmed panel members

Juan Cardenas, UCB Pharma, Spain

Jennifer Chickering, Tisento Therapeutics, USA

Claes Gustafsson, University of Gothenburg, Sweden

Magnus Hansson, Abliva AB, Sweden

Gene Kelly, Stealth BioTherapeutics, USA

Carlos Moraes, University of Miami, USA

17:20 17:35 Sponsor Presentation

Chair: Amel Karaa, Massachusetts General Hospital/Harvard University, USA

An Update on the Zagociguat Phase 2 MELAS Program

Chad Glasser, Tisento Therapeutics, USA

17:35 18:00 Poster pitch talks for even number posters

18:00 19:00 Poster session 2 - even number posters with drinks reception

19:00 Dinner

Bar open (card payments only)

Wednesday 20 March 2024

09:15 Briefing for Keynote, chair, moderator - Auditorium

09:30 10:30 Keynote II

Chair: Rita Horvath, University of Cambridge, UK

Moderator: Robert Pitceathly, University College London, UK

Progress and challenges in AAV gene therapy development for neurological disorders

[Kathrin Meyer, Nationwide Children's Hospital, USA](#)

10:30 11:00 Refreshment break and networking

11:00 Briefing for Session 7 speakers, chair, moderator, committee - Auditorium

11:00 13:00 Session 7: Genomic technologies

Chair: Patrick Chinnery, University of Cambridge, UK

Moderator: Omar Hikmat, Haukeland University Hospital, Norway

11:00 11:30 From Fragmented to Full-Length: A New Era in Human Mitochondrial Genome Sequencing

[Marta Gut, The National Center for Genomic Analysis \(CNAG\), Spain](#)

11:30 12:00 Applying novel technologies to improve the diagnosis of mitochondrial disease

[Robert Taylor, University of Newcastle, UK](#)

12:00 12:30 The role of mitochondrial transcriptional processes in modulating human health and disease

[Alan Hodgkinson, King's College London, UK](#)

12:30 12:45 Constraint as a new tool for identifying deleterious variants and functionally-critical sites in the mtDNA

[Nicole Lake, Yale University, USA](#)

12:45 13:00 Integrated nutrient-sensitized genetic screens predict novel mitochondrial disease genes

[Marcell Harhai, University of Lausanne, Switzerland](#)

13:00 13:10 Closing remarks and prize presentation

Scientific Programme Committee:

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

[Amel Karaa, Massachusetts General Hospital/Harvard University, USA](#)

[Nils-Göran Larsson, Karolinska Institute, Sweden](#)

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

13:10 Lunch and departures

13:50 Coach departures for Stansted and Heathrow airports

14:00 Coach departures for Cambridge train station and city centre