

## Hybrid Conference Programme

19:20

Bar open (card payments only)

Start (GMT)	Finish (GMT)	Presenter details
		mber 2025
12:00	13:00	Registration, lunch and networking
12:45		Briefing for Keynote & Session 1 speakers, microphone runners, chair, moderator & committee - Auditorium
13:00	13:10	Welcome
		Wellcome Connecting Science: Nagehan Ramazanoglu Bahadir, Wellcome Connecting Science, UK Scientific Programme Committee: Amel Karaa - Massachusetts General Hospital/ Harvard University, USA Nils-Goran Larsson - Karolinska Institute, Sweden Michelangelo Mancuso - University of Pisa, Italy Shamima Rahman - University College London, UK
13:10	14:00	Keynote speaker
		Chair: Michelangelo Mancuso - University of Pisa, Italy Moderator: Nils-Goran Larsson - Karolinska Institute, Sweden
		Developing therapeutic approaches by restoring mitochondrial quality control
		Carolyn Sue, The University of New South Wales, Australia
14:00	14:10	Comfort break
14:10	15:40	Session 1: New mechanistic treatments
		Chair: Amel Karaa - Massachusetts General Hospital/ Harvard University, USA  Moderator: Shamima Rahman - University College London, UK
14:10	14:40	Late-onset Thymidine Kinase 2 Deficiency: Clinical Insights, Prognostic Biomarkers, and Pyrimidine Nucleoside Therapy Outcomes
		Christina Dominguez Gonzalez, Hospital 12 de Octubre, Spain
14:40	15:10	Perinatal therapeutic strategies for Surf1-Leigh Syndrome  Dario Brunetti, University of Milan, Italy
15:10	15:25	Ubiquitin-mediated mitophagy regulates the inheritance of mitochondrial DNA mutations  Michele Frison, University of Cambridge, UK
15:25	15:40	Optimising codon sequence and delivery of an AAV9 gene therapy for deoxyguanosine kinase deficiency in brain and liver Arianna Fozzato, University College London, UK
15:40	16:20	Refreshment break and networking
16:05		Briefing for Session 2 speakers, chair & moderator - Auditorium
16:20	17.50	
10.20	17:50	Session 2: Mitochondria as signalling hubs  Chair: Nils-Goran Larsson - Karolinska Institute, Sweden
		Moderator: Michelangelo Mancuso - University of Pisa, Italy
16:20	16:50	Psychobiological Processes in Mitochondrial Diseases  Martin Picard, Columbia University, USA
16:50	17:20	Modulating Mitochondrial Proteostasis: CLPP in Disease Intervention <u>Aleksandra Trifunovic, University of Cologne, Germany</u>
17:20	17:35	Dissecting Mitochondrial and Synaptic Dysregulation in LHON Using Human iPSC-Derived Retinal Ganglion Cells Andrea Cavaliere, Neurological Institute "Carlo Besta", Italy
17:35	17:50	Mitochondiral hubs in pontocerebellae hypoplasia type 6: New insights from patient-derived in vitro models Giorgia Babini, University of Bologna, Italy
17:50	18:20	Poster pitch talks for odd number posters
18:20	19:20	Poster session 1 - odd number posters



Tuesday	11 Nove	ember 2025
09:15		Briefing for Session 3 speakers, chair & moderator - Auditorium
09:30	11:00	Session 3: Role of mitochondria in inflamation and immunity
		Chair: Carolyn Sue, University of South Wales, Sydney, Australia Moderator: Amel Karaa - Massachusetts General Hospital/ Harvard University, USA
09:30	10:00	Innate immunity and inflammation in mitochondrial disease  Phillip West, The Jackson Laboratory, USA
10:00	10:30	Immunological dysfunction in primary mitochondrial disease Shamima Rahman, University College London, UK
10:30	10:45	Delineating the neurodegenerative and inflammatory mechanisms underpinning POLG-related mitochondrial epilepsy  Laura Smith, Newcastle University, UK
10:45	11:00	Investigating inflammatory and metabolic changes in a mouse model of Leigh syndrome following the removal from hypoxia Elizaveta Olkhova, Northumbria University, UK
11:00	11:45	Refreshment break and networking
11:30		Briefing for Session 4 speakers, chair & moderator - Auditorium
11:45	13:15	Session 4: New diagnostic technologies
		Chair: Nils-Goran Larsson - Karolinska Institute, Sweden Moderator: Michelangelo Mancuso - University of Pisa, Italy
11:45	12:15	Metabolic proteins in time and space — Insights from single-cell proteomics  Florian Rosenberger, Karolinska Institute, Sweden
12:15	12:45	Focusing on mitochondria with super-resolution light microscopy Stefan Jakobs, Max Planck Institute for Multidisciplinary Sciences in Göttingen, Germany
12:45	13:00	Correlating "MitoPaint", a novel image-based profiling platform for characterising mitochondrial function, with proteomics to reveal drug-protein functional axes  Sarah Franks, University of Oxford, UK
13:00	13:15	Graphene Windows: Innovating Liquid-Phase Electron Microscopy Methods for Mitochondrial Research  Katie Beirns, Rosalind Franklin Institute, UK
13:15	14:30	Lunch and networking
14:15		Briefing for Session 5 speakers, chair & moderator - Auditorium
14:30	15:45	Session 5: Aspects of mitochondrial biology and disease
		Chair: Michelangelo Mancuso - University of Pisa, Italy Moderator: Shamima Rahman - University College London, UK
14:30	14:45	Ten Years of Next Generation Sequencing: How Genetic Diagnosis and Molecular Epidemiology of Mitochondrial Diseases Have Evolved in Spain  Marcello Bellusci, H.12 de Octubre - CIBERER, Spain
14:45	15:00	Patient perspectives on receiving a diagnosis of primary mitochondrial disease  Katie Waller, The Lily Foundation, UK
15:00	15:15	Development of a targeted approach for introducing single large-scale mtDNA deletions in vivo  Harry Walker, University of Cambridge, UK
15:15	15:30	Ribonucleotide diphosphate reductase small subunit (RnrS) downregulation in Drosophila post-mitotic neuron cells demonstrated the essential role of the enzyme for Central Nervous System activity  Elisa Dell'Anna, University of Bologna, Italy
15:30	15:45	Quantification of mitochondrial complex I deactive/active transition during ischaemia in the murine heart  Katherine Hammond, University of Cambridge, UK
15:45	16:25	Refreshment break and networking
16:25	16:55	Poster pitch talks for even number posters
16:55	17:55	Poster session 2 - even number posters
18:00		Dinner
18:00		Bar open (card payments only)



09:15		Briefing for Session 6 speakers, panel members, chair & moderator - Auditorium
9:30	11:20	Session 6: Clinical trials
		Chair: Shamima Rahman - University College London, UK Moderator: Michelangelo Mancuso - University of Pisa, Italy
09:30	09:50	Clinical Trials and Registries in Rare Neurological Diseases: Lessons, Challenges, and Opportunities  Teresinha Evangelista, The Institute of Myology, France
09:50	10:10	Mitochondrial disease drug development: the saga of sonlicromanol
10:10	10:30	Jan Smeitink, Radboud University, Netherlands  Challenges in development of medicines for in rare neurological diseases  Violeta Stoyanova-Beninska, European Medicines Agency (EMA), Netherlands
10:30	10:50	Nucleic Acid Therapy for Rare Disease - the current status and future perspectives  Haiyan Zhou, University College London, UK
10:50	11:05	Cognitive dissonance: An approach to characterizing and assessing cognitive deficits in primary mitochondrial disease clinical trials Chad Glasser, Tisento Therapeutics, USA (sponsored talk)
11:05	11:35	Panel discussion
		Chair: Amel Karaa - Massachusetts General Hospital/ Harvard University, USA Moderator: Michelangelo Mancuso - University of Pisa, Italy
		Teresinha Evangelista, The Institute of Myology, France
		Jan Smeitink, Radbound University, Netherlands
		Violeta Stoyanova-Beninska. European Medicines Agency (EMA). Netherlands Haiyan Zhou, University College London, UK Chad Glasser, Tisento Therapeutics, USA
11:35	12:10	Refreshment break and networking
11:55		Briefing for Keynote, chair, moderator & committee - Auditorium
12:05	12:55	Keynote speaker
		Chair: Shamima Rahman - University College London, UK
		Moderator: Nils-Goran Larsson - Karolinska Institute, Sweden
		Mitochondria in ischaemia reperfusion injury
		Mike Murphy, University of Cambridge, UK
12:55	13:05	Closing remarks and prize presentation
		Scientific Programme Committee:
		Amel Karaa - Massachusetts General Hospital/ Harvard University, USA
		Nils-Goran Larsson - Karolinska Institute, Sweden Michelangelo Mancuso - University of Pisa, Italy
		Shamima Rahman - University College London, UK
13:05	14:00	Lunch and departures
14:00		Coach departures for Stansted and Heathrow airports
14:15		Coach departures for Cambridge train station and city centre