



UMDF Mitochondrial Medicine 2025 Scientific/Clinical Program

Wednesday, June 18

Wed Morning - Platform Session 1 – North American Mitochondrial Disease Consortium (NAMDC) – *Michio Hirano, MD, Chair – Grand D*

7:00a-6:00p	Registration Open – <i>Grand Foyer Registration</i> 7:00a-8:30a: Continental Breakfast – <i>Grand Foyer</i>
8:00a-8:30a	Welcome and Opening Remarks <i>Philip Yeske, PhD, UMDf Science & Alliance Officer</i> <i>Patient Story: Life with Mitochondrial Disease - The Kallaos Family</i>
8:30a-9:00a	North American Mitochondrial Disease Consortium Impact <i>Michio Hirano, MD, Columbia University Irving Medical Center, New York, NY</i>
9:00a-10:00a	Patient Engagement in NAMDC to Advance the Diagnostic Journey and Disease Understanding <ul style="list-style-type: none"> • Overview of NAMDC Registry and Biorepository - <i>Emanuele Barca, MD, PhD</i> • Mitochondrial Clinical Syndromes – Laying the Groundwork - <i>Valentina Emmanuele, MD, PhD</i> • Mitochondrial Patient Diagnostic Odyssey - <i>JLP Seamus Thompson, PhD</i> • Systematic Studies on the Functional Diagnosis of Mitochondrial Disorders - <i>Johan Van Hove, MD, PhD</i>
10:00a-10:30a	Break – <i>Grand Foyer</i>
10:30a-11:30a	Clinical Trial Readiness, Clinical Studies, and Clinical Trials <ul style="list-style-type: none"> • Mitochondrial Myopathy Outcome Measures - <i>Zarazuela Zolkipli Cunningham, MD</i> • Pyruvate Dehydrogenase Complex Deficiency: Current Clinical Trials and Novel Pharmacological Therapeutics – <i>Jirair K. Bedoyan, MD, PhD</i> • Citrulline Study for MELAS - <i>Fernando Scaglia, MD</i>
11:30a-12:00p	NAMDC Career Enhancement Program <ul style="list-style-type: none"> • <i>Richard Haas, MD; Austin Larson, MD; Zarazuela Zolkipli-Cunningham, MD; Emanuele Barca MD, PhD; and Valentina Emmanuele, MD, PhD</i>
12:00p-12:15p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000017 - <i>Fatima Al Khyeli, MD</i> <i>Title: Assessing Risk for Poor Neurodevelopmental Outcomes in Pediatric Mitochondrial Disease</i>
12:15p-12:30p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000072 - <i>Jean Flickinger, PT, MPT, PCS</i> <i>Title: UMDf study of digital gait and balance sensors characterize fatigue and enhance MM-COAST data in primary mitochondrial disease</i>
12:30p-12:45p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000075 - <i>Stephen C Ekker, PhD</i> <i>Title: Functional Genetic Testing of the Mitochondrial Genome Using Enhanced TALE Base Editors</i>





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Wed Afternoon - Platform Session 2 – Inflammation and Metabolic Diseases – *Fernando Scaglia, MD, Chair – Grand D*

12:45p-2:00p	Lunch – Regency A-D
2:00p-2:30p	Mitochondrial Diabetes in the University of Chicago Monogenic Diabetes Registry <i>Louis H. Philipson, MD, PhD, University of Chicago</i>
2:30p-3:00p	In Vivo Assessment of Diabetes in Primary Mitochondrial Disorders <i>Shana McCormack, MD, MTR, Children's Hospital of Philadelphia (CHOP)</i>
3:00p-3:30p	Mitochondrial Regulation of Thermogenic Fat Activation <i>Jun Wu, PhD, University of Michigan, Life Sciences Institute</i>
3:30p-4:00p	Break – Grand Foyer
4:00p-4:30p	Mitophagy in Obesity and Adipose Inflammation <i>Jon Brestoff, MD, PhD, Washington University of St. Louis, St. Louis, MO</i>
4:30p-5:00p	Mitochondrial Retrograde Signaling in Metabolic Tissues <i>Scott A. Soleimanpour, MD, University of Michigan</i>
5:00p-5:15p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000060 - Wentong Jia, PhD <i>Title: Identification of MXRA8 as a therapeutic target against obesity</i>
5:15p-5:30p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000140 - Norma Frizzell, PhD <i>Title: Exploring anaplerotic therapy in a mouse model of mitochondrial Complex I deficiency</i>
5:30p-7:30p	Break and Poster Sessions <i>Even Numbers Stay at Posters (Non-CME) – Grand AB</i>

Thursday, June 19

Thu Morning - Platform Session 3 - Mitochondria on the Move – *Jonathan R. Brestoff, MD, PhD, MPH, Chair – Grand D*

7:00a-6:00p	Registration Open – Grand Foyer Registration 7:00a-8:30a: Continental Breakfast – Grand Foyer
8:00a-8:30a	Mitochondrial Transfer and Transplantation Enhance Endothelial Engraftment via Mitophagy <i>Juan Melero-Martin, PhD, Boston Children's Hospital / Harvard Medical School, Cambridge, MA</i>
8:30a-9:00a	Release of Cell-free Mitochondria into the Blood: Origins, Functions and Potential Diagnostic Power <i>Alain Thierry, PhD, INSERM/Montpellier University, France</i>





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9:00a-9:30a	Inter-organ Transfer of Mitochondria from Adipocytes to the Heart <i>Clair Crewe, PhD, Washington University St. Louis, St. Louis, MO</i>
9:30a-9:45a	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000032 - Siddhesh Aras, MBBS, PhD <i>Title: Uncovering a linchpin therapeutic target in models of mitochondrial dysfunction</i>
9:45a-10:00a	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000020 - Estefania Nova-Lamperti, PhD <i>Title: Mitochondrial Transfer in Oral Cancer impairs CD4+ T cell responses by promoting cell exhaustion via hypoxic stress</i>
10:00a-10:30a	Break – Grand Foyer
10:30a-11:00a	Tunneling Nanotubes and Mitochondrial Dynamics Within and Between Cells <i>Chiara Zurzolo, MD, PhD, Institut Pasteur, Paris, France</i>
11:00a-11:15a	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000055 - Aaron Bowen, MD, PhD <i>Title: Regional metabolic dynamics in a mouse model of Leigh syndrome</i>
11:15a-11:30a	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000115 - Stella Varnum, Grad Student <i>Title: Mitochondria transfer-based therapies reduce the morbidity and mortality of Leigh syndrome</i>
11:30a-11:45a	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000127- Kyrie Wilson, PhD <i>Title: Development of a Novel Approach to Mitochondrial Gene Therapy: Nanoparticle Mediated Delivery of Intact, Functional Mitochondrial DNA</i>
11:45a-12:00p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000136 - Elizabeth M. McCormick, MS, LCGC <i>Title: Expanding reproductive options for mtDNA disorders in the United States</i>
12:00p-12:15p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000142 - Graeme Preston, PhD <i>Title: PDE5-Inhibitors Increase Coupling Efficiency and Attenuate Hypermetabolism in Primary Mitochondrial Disease</i>
12:15p-12:30p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000148 - Samantha Krysa, PhD <i>Title: Neutrophils enhance defense of cold-induced hypothermia via mitochondria transfer to brown adipocytes</i>





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Thu Morning – Mitochondrial Medicine Society (MMS) – Concurrent Platform – Mary Kay Koenig, MD and Amy Goldstein, MD, Chairs – Regency E-F

7:00a-5:30p	Registration Open – <i>Grand Foyer Registration</i> 7:30a-8:30a: Continental Breakfast – <i>Regency Foyer</i>
8:30a-9:00a	ICU Management with Acidosis Focus <i>Divakar Mithal, MD, PhD, Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL</i>
9:00a-9:30a	Update on Stroke Management in the Mitochondrial Disease Patient <i>Amel Karaa, MD, Massachusetts General Hospital, Boston, MA</i> <i>Amy Goldstein, MD, Children's Hospital of Philadelphia, PA</i>
9:30a-10:00a	Seizing Control: Managing Seizures in Mitochondrial Disease <i>Russell Saneto, DO, PhD, Seattle Children's Hospital, Seattle, WA</i>
10:00a-10:30a	Break – <i>Regency Foyer</i>
10:30a-11:00a	Case Studies - MELAS/POLG <i>Melissa Walker, MD, PhD, Massachusetts General Hospital, Boston, MA</i>
11:00a-12:00p	Navigating Catastrophic Mitochondrial Complications: An Interactive Feedback Session <i>Mary Kay Koenig, MD, UTHealth/McGovern Medical School, Houston, TX</i>
12:00p-12:15p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000083 - Xiaowu Gai, PhD <i>Title: A virtual registry of 9,300 primary mitochondrial disease cases constructed through semi-automated literature mining and expert curation</i>
12:15p-12:30p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000097 - Amy Goldstein, MD <i>Title: Retrospective Natural History Study of POLG Disease in A Mitochondrial Medicine Clinical Research Center</i>
12:30p-2:00p	Lunch – <i>Regency A-D</i>

Thu Afternoon - Platform Session 4 - Mechanisms of Clearing Damaged Mitochondria – Natalie Niemi, PhD, Chair – Grand D

2:00p-2:30p	Mitochondrial Quality Control <i>Åsa Gustafsson, PhD, University of California San Diego</i>
2:30p-3:00p	Mitochondrial Dynamics and Mitochondrial Quality Control <i>David Chan, MD, PhD, California Institute of Technology</i>
3:00p-3:30p	Spatial Regulation of Mitophagy <i>Jonathan Friedman, PhD, UT Southwestern Medical Center, Dallas, TX</i>





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Thu Afternoon - Platform Session 4 - Mechanisms of Clearing Damaged Mitochondria – Natalie Niemi, PhD, Chair – Grand D

3:30p-4:00p	Break – Grand Foyer
4:00p-4:30p	Mitophagy in Health and Disease <i>Martin Graef, PhD, Cornell University, Ithaca, NY</i>
4:30p-4:45p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000026 - Nir Qvit, PhD <i>Title: CVP-350: A Next-Generation Drp1/Fis1 Inhibitor for Targeting Mitochondrial Dysfunction in Cardiovascular Diseases</i>
4:45p-5:00p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000043 - Lia Mayorga, MD, PhD <i>Title: Rewriting nuclear epigenetic scripts in mitochondrial diseases as a strategy for heteroplasmy control</i>
5:00p-5:15p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000046 - Olivia Sniezek-Carney, PhD <i>Title: Evaluating the role of mitophagy and bioenergetic dysfunction in muscle cell differentiation in an iPSC-derived skeletal muscle model of Barth syndrome</i>
5:15p-5:30p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000102 - Shahreen Amin, MD, PhD <i>Title: Mitochondria-lysosome crosstalk alters basal mitophagy in human cells in the neuro-developmental lysosomal disease MPS VII</i>
5:30-5:45p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000051 - Keri-Lyn Kozul, PhD <i>Title: A novel pathogenic mutation of PPTC7 results in metabolic and mitophagy defects characteristic of mitochondrial</i>
5:45-6:00p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000130 - Alexander Stover, MS <i>Title: Impaired bioenergetics in FBXL4-deficient fibroblasts is restored by aminolevulinate and iron (ALA/Fe)</i>
6:00p-8:00p	Poster Sessions - Odd Numbers Stay at Posters - Reception Note: Poster Sessions are Non-CME – Grand AB

Friday, June 20

Fri Morning - Platform Session 5 - Combined Sessions/Clinical Trial Updates – NON-CME

7:00a-5:00p	Registration Open – Grand Foyer Registration 7:30a-9:00a: Continental Breakfast – Regency Foyer
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Friday, June 20

Fri Morning - Platform Session 5 - Combined Sessions/Clinical Trial Updates – NON-CME

9:00a-10:00a	Voices of UMDF and the Big Pitch - A Combined Session for Patient/Family and SciMed Attendees - NON CME – Regency A-D <i>Join us to hear from our Mitochondrial Medicine Community <u>AND</u> don't miss out experiencing the accelerator Big Pitch Presentations!</i>
10:00a-10:30a	Break – Grand Foyer
10:30a-12:30p	Clinical Trial Updates (Non-CME) – Grand D <i>(see mobile app for details on presenters and topics)</i>
12:30p-2:00p	Lunch - Networking by Geographic (for those interested in connecting with attendees in their regions) – Regency A-D

Fri Afternoon - Platform Session 6 – Multi-omics – David J. Pagliarini, PhD, Chair - Grand D

2:00p-2:30p	Systems Biochemistry Approaches to Defining Mitochondrial Protein Function <i>David Pagliarini, PhD, Washington University of St. Louis, St. Louis, MO</i>
2:30p-3:00p	Massively Parallel Single-cell Mitochondrial DNA Genotyping <i>Vijay G. Sankaran, MD, PhD, Boston Children's Hospital/Harvard Medical School, Cambridge, MA</i>
3:00p-3:30p	Multi-omics Discovery Platform for Metabolic Gene Function <i>Eric Gamazon, PhD, MS, Vanderbilt University Medical Center, Nashville, TN</i>
3:30p-4:00p	Break – Grand Foyer
4:00p-4:30p	Integrative Omics Analyses to Improve Diagnostic Rates of Mendelian Disorders <i>Holger Prokisch, PhD, Helmholtz Zentrum München, München, Germany</i>
4:30p-4:45p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000064 - Patrick Forny, MD, PhD <i>Title: Defining Mitochondrial Proteome Regulation via Multi-modal Data Integration</i>
4:45p-5:00p	Abstract Short Talk Presentation (Non-CME) 2025PA-0000000086 - Johan Van Hove, MD, PhD <i>Title: Untargeted proteomics greatly aides the functional diagnosis of mitochondrial aminoacyl-tRNA synthetases (ARS2s)</i>
5:00p-5:15p	Abstract Cash Award Presentations <i>Philip Yeske, PhD, UMDF Science & Alliance Officer</i>





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5:30p-9:30p

UMDF Evening of Energy Reception and Banquet Celebration for ALL Attendees

Saturday, June 21

Masterclass - Up and Coming Clinician – Track 1 – *Michio Hirano, MD, Chair* Primary Mitochondrial Myopathy (PMM) with a Focus on TK2d – *Grand D*

7:30a-2:00p Registration Open – *Grand Foyer Registration*
7:30a-9:00a: Continental Breakfast – *Grand Foyer*

9:00a-9:15a Pre-Program Assessment and Course Overview
Michio Hirano, MD, Columbia University Irving Medical Center, New York, NY

9:15a-9:45a Primary Mitochondrial Myopathy Overview
Fernando Scaglia, MD, Baylor College of Medicine, Houston, TX

9:45a-10:15a Novel Approaches for Achieving a Diagnosis in Mitochondrial Disease Patients
Austin Larson, MD, University of Colorado Anschutz Medical Campus, Aurora, CO

10:15a-10:20a Patient Diagnostic Journey
Patient/Caregiver – Nikki Huggan

10:20a-10:40a Case Study – TK2d
Michio Hirano, MD, Columbia University Irving Medical Center, New York, NY

10:40a-10:45a TK2d Patient – Burden of Disease
Patient – Jared Goettemoeller

10:45a-11:00a Break – *Grand Foyer*

11:00a-11:30a Case Study – Primary Mitochondrial Myopathy
Zarazuela Zolkipli Cunningham, MD, Children's Hospital of Philadelphia, Philadelphia, PA

11:30a-11:35a Primary Mitochondrial Myopathy Patient – Current Treatments
Patient – Megan Leahey

11:35a-12:05p Symptom Management
Michio Hirano, MD, Columbia University Irving Medical Center, New York, NY

12:05p-12:50p Multidisciplinary Management
Amy Goldstein, MD, Children's Hospital of Philadelphia and Amel Karaa, MD, Massachusetts General Hospital, Boston, MA

12:50p-2:30p Lunch Panel Discussion with all Speakers from both Masterclasses – *Grand E*

Masterclass – Early Career Investigators – Track 2 (Non-CME) – *Jonathan Brestoff, MD, PhD, Chair* – *Grand E*

7:30a-2:00p Registration Open – *Grand Foyer Registration*
7:30a-9:00a: Continental Breakfast – *Grand Foyer*





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9:00a-9:15a	Opening with Pre-program Assessment and Course Overview <i>Jonathan Brestoff, MD, PhD</i>
9:15a-9:30a	Faculty Job Search <i>Jonathan Brestoff, MD, PhD and Natalie Niemi, PhD</i>
9:30a-9:45a	Academic Career Success <i>Brett Kaufman, PhD and David Pagliarini, PhD</i>

Saturday, June 21

Masterclass – Early Career Investigators – Track 2 (Non-CME) – Jonathan Brestoff, MD, PhD, Chair – Grand E

9:45a-10:00a	Academic - How to Build Industry Partnerships <i>Bruce Cohen, MD and Douglas Wallace, PhD</i>
10:00a-12:30p	Breakout Discussion Groups
	<p>Session Description - Subject to Change The small group discussions will focus on key topics related to academic and career success in the field of biomedical sciences, providing an interactive and dynamic platform for participants to engage with experts and peers. These discussions will be broken into three timed rounds, allowing attendees to rotate and explore different areas of interest. Each round will be followed by a short break, during which participants or faculty will rotate to a different table(s), ensuring diverse perspectives and the opportunity to engage with different experts and topics. These small group discussions are designed to foster in-depth conversations and provide tailored advice to individuals at various stages of their academic and professional journeys. Below is a brief description of each breakout session.</p> <p>Discussion Group 1: Faculty Job Search Led by Jon Brestoff, MD, PhD, and Natalie Niemi, PhD, this session will guide participants through the nuances of the faculty job search process. This discussion will provide practical tips and strategies for those looking to transition into faculty positions. Participants will have the opportunity to ask questions, share experiences, and gain insights from speakers, and fellow attendees.</p> <p>Discussion Group 2: Academic Career Success Led by Brett Kaufman, PhD and David Pagliarini, PhD, this session will focus on strategies for achieving success in academic careers. Discussions will cover topics such as mentorship, publishing, networking, and navigating the challenges of academic life. Participants will have the opportunity to ask questions, share experiences, and gain insights from Dr. Kaufman, Dr. Pagliarini, and fellow attendees.</p> <p>Discussion Group 3: Academic-Industry Partnerships Led by Bruce Cohen, MD and Douglas Wallace, PhD, this discussion will explore how to build and maintain effective partnerships between academia and industry. Attendees will examine the benefits and challenges of these partnerships, discuss ways to leverage industry collaborations for research advancement, and gain insight into securing industry support for academic projects. Participants will have the opportunity to ask questions, share experiences, and gain insights from speakers, potential Industry representatives, and fellow attendees.</p>





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12:30p-2:30p

Lunch Panel Discussion with all Speakers from both Masterclasses – *Grand E*

