



# Mitochondrial Medicine 2022: Phoenix, AZ

June 8-11, 2022 – Scientific/Clinical Sessions

JW Marriott Phoenix Desert Ridge Resort

*\*Note – Times are Pacific Daylight Time/Mountain Standard Time and subject to change.*

## Day 1: June 8 (Wednesday)

### Morning Platform Session (1) – Platform Title: Directed Gene and Stem Cell Therapy - Scientific Platform (CME)

8:00a **Welcome, Overview and Patient Case** — Brian Harman, President & CEO and Richard Haas, MB, BChir, and Robert K. Naviaux, MD, PhD, Course Co-Chairs

**What Does Mitochondrial Disease Mean to Me** -- Belnap Family, Mito Parent/Patient Perspective

8:30a **Designer DNA Drug Therapy for Neurodegenerative Disease**

Don W. Cleveland, PhD, University of California San Diego School of Medicine, San Diego, CA

9:00a **Reproductive Options for mtDNA Disease**

Robert McFarland, PhD, MRCPCH, Wellcome Centre for Mitochondrial Research, Newcastle University, UK

9:30a *Abstract Presentations (4 slots) 15 minutes each - ([Jump to Abstract Schedule](#))*

**10:30a Break**

11:00a **Directed Gene and Stem Cell Therapy**

Alysson R. Muotri, Ph.D., UCSD School of Medicine, Institute of Genomic Medicine, La Jolla, CA

11:30a *Abstracts Presentations (4 slots) - ([Jump to Abstract to Abstract Schedule](#))*

**12:30p Lunch**

### Afternoon Platform Session (2) – Mitochondrial Medicine Society Platform Session – (CME)

2:00-2:15p **Welcome and MMS Updates**

Amel Karaa, MD, Massachusetts General Hospital, Boston, MA

2:15-2:45p **Management of Acute Emergencies (ICU) in Mitochondrial Disease**

Divakar Mithal, MD, Children's Hospital Chicago, Chicago, IL

2:45-3:15p **Leber's Hereditary Optic Neuropathy (LHON) - Therapy Updates**

Alfredo Sadun, MD, PhD, University of California Los Angeles, Los Angeles, CA

3:15-3:45p **Break**

3:45-4:15p **All Grown Up and Nowhere to Go: The Current State of Adult Metabolic Medicine in the US**

Jessica Gold, Children's Hospital of Philadelphia, Philadelphia, PA

4:15-4:45p **Movement Disorders in Mitochondrial Disease - with a Leigh Syndrome Focus**

Nivedita Thakur, MD, Children's Hospital of Philadelphia, Philadelphia, PA

4:45p *Abstract Presentations (4) - 15 minutes each - ([Jump to Abstract Schedule](#))*

5:45p *Break and Poster Sessions (Non CME)—Even numbers stay at posters*

**7:45p Adjourn**

*A 5-minute question and answer period is included in each of the speaker's allotted time.*

## Day 2: June 9 (Thursday)

### Morning Platform Session (3) – Platform Title: Scientific - Mitochondria in Space (CME)

- 8:00a **Welcome and Overview and Patient Perspective** - Robert K. Naviaux, MD, PhD, Course Co-Chair and Jana Stoudemire, Mito Parent and UMDF Trustee
- 8:15a **Mitochondrial Responses to Long-Term Space Flight, the International Space Station Experience**  
Hemal H. Patel, PhD, University of California San Diego, California
- 8:45a **Space-Flight Associated Neuro-ocular Syndromes**  
Tyson Brunstetter, OD, PhD, NASA Johnson Space Center, Houston, TX
- 9:30a *Break*
- 10:00a **Cell-free DNA (cfDNA) and Exosome Profiling from a Year-long Human Spaceflight Reveals Circulating Biomarkers**  
Daniela Bezdan, Institute of Medical Genetics and Applied Genomics, University of Tübingen, Tübingen, Germany
- 10:30a **Mitochondria Dysfunction as the Universal Driver of Space Flight Across All Species**  
Afshin Beheshti, PhD, KBR at NASA Ames Research Center, Mountain View, CA
- 11:00a **Roundtable Question and Answer Session with the Speakers**
- 12:00p *Abstract Presentations (2 slots) - ([Jump to Abstract Schedule](#))*
- 12:30p Lunch**

### Afternoon Platform Session (4) – Platform Title: Clinical Trial Readiness and the Voice of the Patient Industry/Clinical Platform – NON CME

- 1:45p **Overview of Session - What to Expect** - Philip Yeske, PhD
- 2:00p **Industry/Clinical Session – New Objective Outcome Measures of Myopathy for Primary Mitochondrial Disease Clinical Trials** - Zarazuela Zolkipli-Cunningham, MBChB, MRCP, Children's Hospital of Philadelphia, Philadelphia, PA
- 2:30p **Clinical Trial Readiness - Regulatory Landscape**
- 2:30p-3:00p **The Regulatory Perspective on Rare Disease Drug Development**  
Anna Choe, MD, MPH, Food and Drug Administration
- 3:00p-3:30p **Role of the Rare Disease Cures Accelerator- Data and Analytics Platform in Rare Disease Drug Development**  
Alexandre Bétourné, PhD, PharmD, Critical Path Institute
- 3:30p Break**
- 4:00p **Voice of the Patient - Ask the Mito Patient Session**  
*In our quest to ensure mitochondrial disease drug development advances based on valuable patient perspectives, this session will not only capture the patient voice but it will give clinicians, researchers, and industry an opportunity to ask questions related to clinical trial readiness.*  
*Tentative Make-up of Patient/Caregiver Panelists: Ped/Caregiver Leigh, Adult MELAS, LHON adult patient, POLG patient, CPEO and/or KSS*  
**Unable to Attend? [Click Here for Virtual Option](#)**

- 4:00p **Welcome & What to Expect**, Brian Harman, President & CEO, UMDF  
4:05p **FDA Listening Session - TK2d Summary Readout**, Michio Hirano, MD  
4:15p **Patient/Caregiver Journeys** - 5 panelists will provide a 3 min journey introduction - 15 mins  
4:30p **Symptoms** - with Polling and Q&A - 10min  
4:40p **Burden of Disease** - with Polling and Q&A - 10min  
4:50p **Current Treatments** - with Polling and Q&A - 10min  
5:00p **Idealized Treatments and Meaningful Quality of Life Improvements** - 10min  
5:10p **Closing Comments**  
5:15p **Adjourn**

5:00p **Pre-Clinical Model Systems - Abstract Presentations (4 slots)** - ([Jump to Abstract Schedule](#))

6:00p **NEW Special Poster Sessions and Meet/Greet Reception - Heavy Hors d'oeuvres and Cash Bar** - Odd numbers stay at posters  
This will be a special kick-off reception with the entire community.

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### Day 3: June 10 (Friday)

**Morning Platform Session (5) – Platform Title: State of Mitochondrial Medicine (Scientific, Clinicians, Patients, Families, etc). NON CME**

8:00a **Welcome: UMDF and the Mitochondrial Medicine Community - Sonoran FG**  
Speaker: Brian Harman, UMDF President & CEO, Pittsburgh, PA

8:20a **UMDF Impact**  
Philip Yeske, PhD, UMDF Science & Alliance Officer, Pittsburgh, PA

8:45a **The Big Pitch: UMDF accelerators Research Grant Program Presentations**

9:45a **Break and Special Networking**

10:15a **Clinical Trial Breakout Session Overview - Saguaro East West**  
Philip Yeske, PhD

10:20a **Current Rare Disease Landscape for Clinical Trials**  
Frank Sasinowski, MS, MPH, JD, Hyman, Phelps and McNamara

10:45a **Clinical Trial Survey Process and Summary**  
Amel Karaa, MD

11:00a **Industry Focused Breakout with Sci/Med and Patients/Caregivers**

11:30a **Clinician Focused Breakout with Sci/Med and Patients/Caregivers**

12:00p **Lunch - Grand Sonoran FG**

1:00p **Initial Summary of Clinical Trial Readiness Breakouts and Goal - Grand Sonoran FG**

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**Afternoon Platform Session (6) – Platform Title: Clinical Trial Related Sessions – NON CME**

2:00p **Clinical Trial Updates with Discussion Panels - Saguaro East West**

2:00p **Clinical Trial Panel 1 – Moderator: Richard Haas, MB, BChir**  
GenSight Biologics, Minovia Therapeutics, PTC Therapeutics,  
University of Florida/Saol Therapeutics

- 2:30p Clinical Trial Panel 2 – Moderator: Michio Hirano, MD**  
Abliva, Baylor College of Medicine, Cycleron Therapeutics, Khondrion
- 3:00p Break**
- 3:30p Clinical Trial Panel 3 – Moderator: Amel Karaa, MD**  
Astellas Pharma, Reneo Pharmaceuticals, Stealth BioTherapeutics, Zogenix now part of UCB
- 4:00p UMDF Supported Infrastructure Panel Discussions – Moderator: Philip Yeske, PhD**  
North American Mitochondrial Disease Consortium (NAMDC); Mitochondrial Disease Sequence Data Resource Consortium (MSeqDR); Mitochondrial Care Network (MCN); and TreatMito
- 4:45p MMS/MRS Abstract Awards and Gateway Grant Awards - Saguaro East West**  
Philip Yeske, PhD, UMDF Science & Alliance Officer
- 5:00p Adjourn**
- 5:30p Reception and Cash Bar**
- 6:30p An Evening of Energy Banquet Celebration for Scientific/Clinical and all Patient/Family Attendees - music, accelerator vote, and Awards Ceremony - Sonoran F-G**

## Day 4: June 11 (Saturday)

### Morning Platform Session (7) – Platform Title: Autism Inflammation and Mitochondria - CME

- 8:00a Autism and Mitochondria—2010 to 2022**  
Robert K. Naviaux, MD, PhD, University of California San Diego, CA
- 8:30a Early Life Stress and Brain Mitochondrial Dysfunction**  
Pat Levitt, PhD, Children’s Hospital Los Angeles, Los Angeles, CA
- 9:00a Early Intervention – Autism Spectrum Disorders**  
Rebecca Grzadzinski, PhD, University of North Carolina, Chapel Hill, NC
- 9:30a Neuroimmunology and Autoimmunity in Autism Spectrum Disorders**  
Judy Van de Water, PhD, University of California Davis, Davis, CA
- 10:00a Break**
- 10:30a Multiomic Features of the Microbiome and Autism**  
Brittany Needham, PhD, California Institute of Technology, Los Angeles, CA
- 11:00a Electrophysiologic Insights to Neurodevelopment in Autism Spectrum Disorders**  
Abigail Dickinson, PhD, University of California Los Angeles, Los Angeles, CA
- 11:30a Abstract Presentations (3) 15 minutes each—Autism and Neuroinflammation - ([Jump to Abstract Schedule](#))**
- 12:15p Autism Q&A Speakers Round Table**
- 1:00p Closing Remarks and Adjourn – Lunch Provided**  
Richard Haas, MB, BChir, and Robert K. Naviaux, MD, PhD, Course Co-Chairs

## UMDF Mitochondrial Medicine 2022: Phoenix, AZ

### Oral Presentations – Abstracts

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Wednesday, June 8, 2022			
Platform Session #1: <i>Directed Gene and Stem Cell Therapy</i>			
Time	Abstract#	Presenter	Title
9:30a	2022PA-0000000011	Simon Johnson	Leukocytes mediate disease pathogenesis in the Ndufs4(KO) mouse model of Leigh syndrome
9:45a	2022PA-0000000039	Yudong Wang	Restoration of interaction between FAO and ETC proteins in VLCAD and VLCAD-deficient mice mitochondria by addition of recombinant VLCAD
10:00a	2022PA-0000000048	Valeria Di Leo	The effects of resistant exercise training on mitochondrial myopathy patients
10:15a	2022PA-0000000055	Carly Truong	Safe-harbor Allotopic Expression of Mitochondrial ATP8 gene from the Nucleus In Vivo
11:30a	2022PA-0000000094	Sampath Rangasamy	Recurrent Splice Site mutation in Mitochondrial Methionyl-tRNA Formyl Transferase (MTFMT): From Models to Therapy
11:45a	2022PA-0000000027	Anne Chiaramello	Multi-omics profiling and mitochondrial energy metabolism investigations of the ultra-rare MELAS m.14453 G>A variant
12:00p  CME Program Ends for the morning.	2022PA-0000000037	Elizabeth M. McCormick	Facilitating community-based genomic data analysis in primary mitochondrial disease: Collaboration of the Mitochondrial Disease Sequence Data Resource (MSeqDR) and mitoSHARE registry to support future genomic research discoveries.
NON CME 12:15p	2022PA-0000000041	Wendy Shoop	ARCUS Gene Editing to Eliminate MELAS-associated m.3243A>G Mutant Mitochondrial DNA

Platform Session #2: <i>Mitochondrial Medicine Society Platform Session</i>			
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Time	Abstract#	Presenter	Title
4:45p	2022PA-0000000014	Isabel Lopez Sanchez	Establishing Risk of Vision Loss in Leber Hereditary Optic Neuropathy
5:00p	2022PA-0000000089	Renkui Bai	Mitochondrial Diseases Are Common in NICU Patients: Experience from Rapid Exome Sequencing for 1935 infants
5:15p	2022PA-0000000092	Amy Goldstein	Improving Acute Care for Mitochondrial Disease: Development of a Publicly Available Clinical Care Pathway
5:30p	2022PA-0000000023	Eliza Gordon-Lipkin	Side effect profiles of the COVID-19 Vaccine Amongst Patients with Mitochondrial Disease
Thursday, June 9, 2022			
Platform Session #3: <i>Mitochondria in Space and Clinical Trial Readiness</i>			
Time	Abstract#	Presenter	Title
12:00p	2022PA-0000000108	Zahra Tara	Exploiting mitophagy to develop therapies for OPA1 disease in Worm with validation in zebrafish and human fibroblast cell line translational models.
12:15p	2022PA-0000000020	Derek P. Narenda	OMA1 mediates local and global stress responses against protein misfolding in CHCHD10 mitochondrial myopathy
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## Thursday, June 9, 2022

### NON CME - Platform Session #4: *Pre-Clinical Model Systems*

<b>5:00p</b>	2022PA-0000000044	Meghan Eller	Development of a Novel ECHS1 Deficiency Mouse Model
<b>5:15p</b>	2022PA-0000000082	Suraiya Haroon	Single large-scale mitochondrial deletion (SLSMD) disease model in <i>C. elegans</i> demonstrates increased UPRmt response phenotype despite low heteroplasmy levels
<b>5:30p</b>	2022PA-0000000057	Matthew G. Baile	Frataxin (FXN) promotes the Expression of Neuronal Differentiation Markers: Effect of FXN Supplementation in a Neurodegeneration Model
<b>5:45p</b>	2022PA-0000000060	Manuela Lavorato	Dichloroacetate improves survival, neuromuscular function, and mitochondrial physiology and morphology in FBXL4 disease preclinical <i>C. elegans</i> , zebrafish, and human fibroblast models

## Saturday, June 11, 2022

### Platform Session #7: *Autism Inflammation and Mitochondria*

Time	Abstract#	Presenter	Title
<b>11:30a</b>	2022PA-0000000040	Patrick M. Schaefer	A common mtDNA variant on an unorthodox mtDNA background results in connective tissue disorder
<b>11:45a</b>	2022PA-0000000059	Alison Lane	Unique Bioenergetic Profiles of Children with Autism Spectrum Disorder with and without Neurodevelopmental Regression
<b>12:00p</b>	2022PA-0000000077	Brianna A. Werner	Mitochondrial Respiration Alterations in Epileptic Foci from Fresh Human Brain