

Nutritional Interventions in Primary Mitochondrial Disorders: Developing an Evidence Base



December 2-3, 2014 | Rockville, Maryland | Neuroscience Center Bldg.

AGENDA

Workshop Goals: Explore the use of nutritional interventions, including dietary supplements, in primary mitochondrial disorders; identify gaps in knowledge; identify research opportunities; develop a research agenda to promote an evidence base for the use of nutritional interventions in primary mitochondrial disorders; and forge collaborations among researchers, clinicians, patient advocacy, and Federal partners.

DAY 1 — TUESDAY, DECEMBER 2

- 7:00 A.M. REGISTRATION**
- 8:00 A.M. Welcome and Introductions**
James Anderson, M.D., Ph.D. — Division of Program Coordination, Planning, and Strategic Initiatives, National Institutes of Health (NIH)
Paul Coates, Ph.D. — Office of Dietary Supplements (ODS), NIH
Kathryn Camp, M.S., R.D., C.S.P. — ODS, NIH
- 8:20 A.M. Session 1. Setting the Stage**
Moderator: *Phil Yeske, Ph.D.* — United Mitochondrial Disease Foundation (UMDF)
- 8:30 A.M. Regulatory Definitions in the United States**
Andrew Mulberg, M.D. — U.S. Food and Drug Administration (FDA)
Shawne Suggs-Anderson, M.S., R.D. — FDA
- 9:30 A.M. Global Efforts**
Amy Goldstein, M.D. — Children's Hospital of Pittsburgh, Mitochondrial Medicine Society
- 10:00 A.M. Questions and Answers (Q&A)**
- 10:10 A.M. BREAK**
- 10:25 A.M. Session 2. Primary Mitochondrial Disorders (PMD)**
Moderator: *Katrina Gwinn, M.D.* — National Institute of Neurological Disorders and Stroke (NINDS), NIH
- 10:30 A.M. MtDNA-Related Disorders**
Michio Hirano, M.D. — Columbia University
- 11:00 A.M. Mendelian Mitochondrial Diseases**
Gregory Enns, M.D. — Stanford University
- 11:30 A.M. Genetics of PMD; Genotype/Phenotype Correlations**
Robert McFarland, M.A., M.B.B.S., Ph.D., M.R.C.P., M.R.C.P.C.H. — Newcastle University



- 12:00 P.M. Q&A**
- 12:15 P.M. LUNCH (ON YOUR OWN)**
- 1:00 P.M. Session 3. How Nutritional Interventions are Used in PMD**
Moderator: *Kathryn Camp, M.S., R.D., C.S.P.* — ODS, NIH
- 1:10 P.M. Nutritional Interventions in PMD; Which Ones are Used and for What Purpose**
Bruce H. Cohen, M.D. — Akron Children's Hospital
Kathryn Camp, M.S., R.D., C.S.P. — ODS, NIH
- 1:40 P.M. Practice Patterns of Physicians in North America**
Sumit Parikh, M.D. — Cleveland Clinic
- 2:00 P.M. North American Mitochondrial Disease Consortium Patient Survey on Dietary Supplement Use**
Amel Karaa, M.D. — Massachusetts General Hospital
- 2:20 P.M. Q&A**
- 2:30 P.M. BREAK**
- 2:45 P.M. Session 4. Challenges and Barriers to Dietary Supplement Use in PMD**
Moderator: *Gregory Enns, M.D.* — Stanford University
- 2:50 P.M. Clinical Trials in Mitochondrial Disease—It is Not Your Father's Oldsmobile: Endpoints, Biomarkers, and Other Barriers**
Bruce H. Cohen, M.D. — Akron Children's Hospital
- 3:30 P.M. Scientific Evidence versus Clinical Practice**
Jerry Vockley, M.D., Ph.D. — University of Pittsburgh
- 4:00 P.M. Q&A**
- 4:15 P.M. Session 5. Perspectives from Patient Advocacy, Industry, and Professional Organizations Panel Discussion:**
Moderator: *Rashmi Gopal-Srivastava, M.S, Ph.D.* — Office of Rare Diseases Research (ORDR), National Center for Advancing Translational Sciences (NCATS), NIH
- Patient Advocacy**
- *Chuck Mohan* — UMDF
 - *Christine Cox, J.D.* — MitoAction
- Industry**
- *Tricia Rutherford* — Vitaflo International Limited
 - *Steven Yannicelli, Ph.D., R.D.* — Nutricia North America

Professional Organizations

- *Jerry Vockley, M.D., Ph.D.* — American College of Medical Genetics and Society for Inherited Metabolic Disorders
- *Erin L. MacLeod, Ph.D., R.D., L.D.* — Genetic Metabolic Dietitians International

5:30 P.M. **ADJOURN**

DAY 2 — WEDNESDAY, DECEMBER 3

8:00 A.M. **OPENING REMARKS**

8:10 A.M. **Session 6. New Technologies and Omic Approaches to Diagnosis, Treatment, and Understanding Mechanisms of Action**

Moderator: *Robert McFarland, M.A., M.B.B.S., Ph.D., M.R.C.P., M.R.C.P.C.H.* — Newcastle University

8:15 A.M. **Integrating the Transcriptome, Metabolome, and Mitochondrial Physiome to Clarify Disease Mechanisms and Treatment Effects in Mitochondrial Respiratory Chain Disease**

Marni Falk, M.D. — University of Pennsylvania

8:45 A.M. **Metabolic Profiling of Mitochondrial Disease**

Vamsi Mootha, M.D. — Massachusetts General Hospital

9:15 A.M. **Evidenced-Based Approaches: High Throughput Mitofunctional Screening of Single Molecule and Herbal Supplements**

Gino Cortopassi, Ph.D. — University of California, Davis

9:45 A.M. **The Gut Microbiome Metabolism of Carnitine and Implications for Mitochondrial Disorders**

Wilson Tang, M.D. — Cleveland Clinic

10:15 A.M. **Q&A**

10:30 A.M. **BREAK**

10:45 A.M. **Patient – Product Safety: A Case Study**

Freddie Ann Hoffman, M.D. — Heterogeneity, LLC

11:15 A.M. **Session 7. Research Opportunities and Resources**

Moderator: *Danuta Krotoski, Ph.D.* — Eunice Kennedy Shriver National Institute of Child Health and Human Development (NICHD), NIH

Summary of March 2012 NIH Mitochondrial Meeting

Danuta Krotoski, Ph.D. — NICHD, NIH

11:30 A.M. **Development of Common Data Elements for Mitochondrial Disorders**

Joanne Odenkirchen, M.P.H. — NINDS, NIH

- 11:45 A.M. Outcome Measures**
Example: A New Approach: The EPI-743 Trial
Lynne Wolfe, M.S., C.R.N.P. — National Human Genome Research Institute (NHGRI), NIH
- 12:00 P.M. Perspectives from Government, Patient Advocacy, Industry, Foundations Panel Discussion**
- U.S. Federal Government**
Andrew Mulberg, M.D. — FDA
Katrina Gwinn, M.D. — NINDS, NIH
- United Kingdom**
Grainne Gorman, M.D. — Newcastle University
- Patient Advocacy**
Phil Yeske, Ph.D. — UMDF
- Industry**
Matt Klein, M.D. — Edison
- Foundation**
Stephen Hersh, M.D. — The J. Willard and Alice S. Marriott Foundation
- 12:45 P.M. LUNCH (ON YOUR OWN)**
- 1:30 P.M. Session 8. Developing a Research Agenda**
Moderator: *Melissa Parisi, M.D., Ph.D.* — NICHD, NIH

All participants
- 2:45 P.M. Next Steps**
- 3:00 P.M. ADJOURN**