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**
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**

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**