

# The United Mitochondrial Disease Foundation Scientific Program Preliminary Draft

## Mitochondrial Medicine 2012 *From Genomics and Systems Biology to Translation*

June 13-15, 2012 – Scientific Sessions  
June 16, 2012 – Special Emphasis Sessions  
June 15-16, 2012 – Patient/Family Sessions  
June 14, 2012 – UMDF Day on the Hill Visits

**Bethesda Marriott, Bethesda, MD**

[Day One](#)

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### Day 1: June 13 (Wednesday)

#### **Morning Platform Session (1) – Platform Title: Genomics of Mitochondrial Biology**

- 8:00 a.m. Welcome  
Topic Title: Identification of the Calcium Uniporter via Comparative Genomics  
Speaker: *Vamsi Mootha, MD, Course Chair*
- 8:45 a.m. Topic Title: Genomics vs. Proteomics: Two Views of Mitochondrial Evolution  
Speaker Name: *Michael W. Gray, PhD*
- 9:15 a.m. Topic Title: Remaining Relevant: The Minimalist Mitochondrion of Malaria Parasite  
Speaker Name: *Akhil B. Vaidya, PhD*
- 9:45 a.m. Break**
- 10:15 a.m. *Abstract Presentations (7)*
- 12:00 p.m. Lunch**

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#### **Afternoon Platform Session (2) – Platform Title: Mitochondrial Proteomics**

- 2:00 p.m. Topic Title: *Omics Approaches Reveal Novel Tools for Mitochondrial Disease Diagnosis*  
Speaker Name: *Anu Suomalainen Wartiovaara, MD, PhD*
- 2:30 p.m. Topic Title: *Proteomic Insights into Mitochondrial Form and Function*  
Speaker Name: *Dave Pagliarini, PhD*
- 3:00 p.m. Topic Title: Protein Modifications in the Mitochondria – Driving Heart Disease  
Speaker Name: *Jennifer van Eyk, PhD*
- 3:30 p.m. *Abstract Presentations (3)*
- 4:15 p.m. Break & Poster Sessions (non cme) - Even Numbers Stay at Posters**
- 6:30 p.m. Adjourn**

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

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## Day 2: June 14 (Thursday)

### **Morning Platform Session (3) - Chemical Biology and Metabolomics**

- 8:00 a.m. Topic Title: Redox Biomarkers in Mitochondrial Disease  
*Speaker Name: Gregory M. Enns, MB, ChB*
- 8:30 a.m. Topic Title: Treating Regulatory Signaling Network Changes that Cause the Metabolic Sequelae of Mitochondrial Disease  
*Speaker Name: Marni Falk, MD*
- 9:00 a.m. Topic Title: TBA  
*Speaker Name: Joshua Rabinowitz, MD, PhD*
- 9:30 a.m. Topic Title: The Versatility of Mitochondrial Metabolism in Tumor Cell Growth  
*Speaker Name: Ralph J. DeBerardinis, MD, PhD*

#### **10:00 a.m. Break**

10:30 a.m. *Abstract Presentations (6)*

#### **12:00 p.m. Lunch**

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### **Afternoon Platform Session (4) – Platform Title: Systems Biology and Dynamics**

- 2:00 p.m. Topic Title: The Systems Biology of the Cardiac Mitochondrion  
*Speaker Name: Robert S. Balaban, PhD*
- 2:30 p.m. Topic Title: Genome Technology for Mitochondrial Disease  
*Speaker Name: Curt Scharfe, MD, PhD*
- 3:00 p.m. *Abstract Presentations (5)*
- 3:30pm to 6:30pm** **Special Clinical Director's Workshop** - this session is a new opportunity for clinicians, in an open discussion format, to work on day-to-day challenges faced by clinicians in their groups (patient care and systems related). (NON CME)
- 5:30 p.m. Break and Posters (non cme) - Odd Numbers Stay at Posters Reception and Cash Bar**
- 7:30 p.m. Adjourn**

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

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## Day 3: June 15 (Friday)

### **Morning Platform Session (5) – Next-Generation Sequencing of Mitochondrial Disease**

- 8:00 a.m. Topic Title: MitoExome Sequencing of Children with Mitochondrial Disease  
*Speaker Name: David Thorburn, PhD*
- 8:30 a.m. Topic Title: Challenges of Bringing Next Generation Sequencing Technologies to CLIA/CAP Certified Clinical Laboratories  
*Speaker Name: Lee Jun C. Wong, PhD*
- 9:00 a.m. Topic Title: Exome Sequencing of Mitochondrial Disorders  
*Speaker Name: Eric A. Shoubridge, PhD*
- 9:30 a.m. **Abstract Presentations (2)**
- 10:00 a.m. Break**
- 10:30 a.m. Topic Title: Diagnostic Efficacy of Targeted Sequencing in Infantile Versus Adult Mitochondrial Disease  
*Speaker Name: Sarah Calvo, PhD*
- 11:00 a.m. *Abstract Presentations (5)*
- 12:15 p.m. Lunch**

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### **Afternoon Platform Session (6) – Translational Mitochondrial Medicine**

- 2:00 p.m. Topic Title: Monitoring Disease: the Newcastle Approach  
*Speaker Name: Patrick Chinnery, PhD, FRCP*
- 2:30 p.m. Topic Title: The Mitochondriopathy of Methylmalonic Acidemia :  
Definition and Therapeutic Approaches  
*Speaker Name: Charles P. Venditti, MD, PhD*
- 3:00 p.m. Topic Title: Disease Severity Scores and Outcome Measures in  
Mitochondrial Disease  
*Speaker Name: Jan Smeitink, MD, PhD*
- 3:30 p.m. Break**
- 3:45 p.m. Topic Title: NAMDC  
*Speaker Name: Michio Hirano, MD*
- 4:15 p.m. UMDF 2012 Funded Grant Projects
- 5:30 p.m. Adjourn**
- 6:15 p.m. Reception**
- 7:00 p.m. Friday Night Banquet and Awards Ceremony**  
**Keynote Address: William Gahl, MD, PhD**

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

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### **Day 4: June 16 (Saturday)**

#### **Platform Session (7) - Clinical Algorithms, Biochemical Testing, Exercise Dx**

- 8:00 a.m. Topic Title: Respiratory Chain Testing  
*Speaker Name: Charles Hoppel, MD*
- 8:30 a.m. Topic Title: Biochemical Profiling in Mitochondrial Disease  
*Speaker Name: Richard Haas, MB, BChir*
- 9:00 a.m. Topic Title: Exercise Diagnosis  
*Speaker Name: Ron Haller, MD*
- 9:30 a.m. Topic Title: Oxidative Phosphorylation Disorders: from Bench to  
Bedside  
*Speaker Name: Jan Smeitink, MD, PhD*
- 10:00 a.m. Break**
- 10:30 a.m. Topic Title: Criteria for Diagnosis of Mitochondrial Disease in the Era of  
Next Generation Sequencing  
*Speaker Name: David Thorburn, PhD*
- 11:00 a.m. Topic Title: New Treatments for Mitochondrial Diseases?  
*Speaker Name: Patrick Chinnery, PhD, FRCP*
- 11:30 a.m. Panel Discussion - How to Incorporate Next Generation Profiles  
into Routine Clinical Diagnosis**  
**Moderator: Vamsi Mootha**  
**Panel: Chuck Hoppel, Richard Haas, Ron Haller, Jan Smeitink, Eric  
Shoubridge, David Thorburn, Patrick Chinnery, Anu Wartiovaara,  
and Lee Jun Wong**
- 1:30 p.m. Adjournment**

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

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