Program for SIMD Annual Meeting

Renaissance Nashville Hotel (RNH) and Nashville Convention Center (NCC)
Nashville, Tennessee

Saturday, March 24, 2007

3:00 PM–6:00 PM  
Registration—Renaissance Hotel—Main Lobby

Sunday, March 25, 2007

10:00 AM–6:00 PM  
Registration—Renaissance Hotel—Main Lobby

12:00 PM–6:00 PM  
Exhibit and poster set up—NCC—East Exhibit Hall—Level One

3:00 PM  
Hotel Check-In—Renaissance Hotel

1:30 PM–5:00 PM  
ACMG-SIMD Joint Symposium—Nashville Convention Center (NCC)—Ballroom 206
Expanded Newborn Screening: How are We Dealing with Diagnostic Dilemmas and Counseling? What’s New on the Horizon?
Moderators: Robert Steiner, MD, Oregon Health & Science University
Georgirene D. Vladutiu, PhD, The State University of New York at Buffalo

1:30 PM–2:00 PM  
Confirmatory Newborn Screening Diagnostic Protocols
Dietrich Matern, MD
Mayo Clinic

2:00 PM–2:30 PM  
False Positives and False Negatives: Impact on Physicians and Families
Susan Waisbren, MD
Children’s Hospital, Boston

2:30 PM–3:00 PM  
The Next Generation of Newborn Genetic Testing: Severe Combined Immune Deficiency
Jennifer M. Puck, MD
University of California, San Francisco

3:00 PM–3:30 PM  
Newborn Screening for Adrenoleukodystrophy: Implications for Therapy
Ann Moser, BA
Kennedy Krieger Institute, Baltimore

3:30 PM–4:00 PM  
Coffee Break

4:00 PM–5:00 PM  
Positive Newborn Screens: Challenging Cases
Moderator: Carol L. Greene, MD, University of Maryland
Panel Members:
Barbara K. Burton, MD, Children’s Memorial Hospital, Chicago, IL
Monday, March 26, 2007

7:00 AM–8:15 AM  
**Breakfast—Renaissance Nashville Hotel—East Grand Ballroom Lobby Level**

8:00 AM–12:00 Noon  
**Registration—outside Nashville Convention Center Ballrooms 204-206**

8:00 AM–12:15 PM  
**NCC Ballrooms 204-206—Level Two**  
*Session 1: The Latest Updates on Inborn Errors of Metabolism*  
**Moderator: Nicola Longo, MD, PhD**  
*University of Utah*

8:00 AM–8:45 AM  
Inborn Errors: Past, Present and Future  
**David Valle, MD**  
*Johns Hopkins University*

8:45 AM–9:30 AM  
A Novel, Palmitoyl-Protein Desaturase Function for the Batten Disease Protein  
**Michael Bennett, PhD**  
*The Children's Hospital of Philadelphia*

9:30 AM–10:15 AM  
Expanded DNA Diagnosis of Mitochondrial Disorders. Nuclear Genes  
**Lee-Jun Wong, PhD**  
*Baylor College of Medicine*

10:15 AM–10:45 AM  
**Coffee Break—NCC East Exhibit Hall—Level One**

10:45 AM–11:30 AM  
CPS I Polymorphisms and the Candidate Disease Approach to Research  
**Marshall Summar, MD**  
*Vanderbilt University Medical Center*

11:30 AM–12:15 PM  
New Inborn Errors in the Pentose Phosphate Pathway Affecting Polyol Metabolism: Transaldolase and Ribose-5-Phosphate Isomerase Deficiencies  
**Cornelis Jakobs, MD**  
*VU University Medical Center, Amsterdam*

12:15 PM–1:00 PM  
**Lunch—Renaissance Nashville Hotel—East Grand Ballroom Lobby Level**  
(for paid registrants only)—must pre-register—ticket required

1:00 PM–1:45 PM  
The Third Robert Guthrie Memorial Lecture—*RNH East Grand Ballroom*  
Who is Afraid of MS/MS? Myths and Challenges of Expanded Newborn Screening  
**Piero Rinaldo, MD, PhD**  
*Mayo Clinic*

2:00 PM–5:30 PM  
**Session II: Contributed Papers—NCC Ballrooms 204-206—Level Two**  
Presentations from Travel Award Winners  
**Moderator: William Gahl, MD, PhD**  
*NIH, National Human Genome Research Institute*

2:00 PM–2:15 PM  
Exploring Mucolipidosis II and III  
**Sara S. Cathey, MD**  
*Greenwood Genetics Center*
2:15 PM–2:30 PM  Cobalamin C Disease Identified By Expanded Newborn Screening: The California Experience  
Kristina Cusmano-Ozog, MD  
Stanford University

2:30 PM–2:45 PM  Citrin Deficiency: Apparent Founder Mutations in the French Canadian Population  
David Dimmock, MD  
Baylor College of Medicine

2:45 PM–3:00 PM  Metabolic Pathway Expression Profiling in C. Elegans Mitochondrial Respiratory Chain Mutants  
Marni Falk, MD  
The Children's Hospital of Philadelphia

3:00 PM–3:15 PM  Expression and Characterization of Mutations in Very Long-Chain Acyl-CoA Dehydrogenase Using a Prokaryotic System  
Eric Goetzman, PhD  
Children's Hospital of Pittsburgh

3:15 PM–3:30 PM  A Possible New Disorder of Cholesterol Biosynthesis Involving the 4α-Methylsterol-4-Demethylase Complex  
Miao He, PhD  
Children's Hospital of Pittsburgh

3:30 PM–4:00 PM  Coffee Break—NCC East Exhibit Hall—Level One

4:00 PM–4:15 PM  N-Acetylmannosamine Treatment Rescues GNE Knock-in Mice From Severe Neonatal Glomerular Hematuria and Podocytopathy. Insights for Hereditary Inclusion Body Myopathy  
Enriko D. Klootwijk, PhD  
NIH, National Human Genome Research Institute

4:15 PM–4:30 PM  Acute Presentation of β-Ketothiolase Deficiency: An Illustrative Case for Glucose Administration in Pediatric Advanced Life Support  
Linda M. Lukose, MD  
NIH, National Human Genome Research Institute

4:30 PM–4:45 PM  Multidisciplinary Study of in Utero Presentation of Fetal GM1 Gangliosidosis at 17 Weeks Gestation  
Susan Pattison, CCMG Fellow  
McMaster University

4:45 PM–5:00 PM  Oxidative and Endoplasmic Reticulum Stress In Novel Isogenic Cell Models For Classic Galactosemia  
Manshu Tang  
The Dr. John T. Macdonald Foundation Center for Medical Genetics

5:00 PM–5:15 PM  The Effect of Phenylalanine and Tyrosine Intake on Tetrahydrobiopterin (BH4) Response in Phenylketonuria (PKU)  
Andrea Wierenga, PhD  
University of Miami

5:15 PM–5:30 PM  Development of a High-Throughput Assay for the Identification of Small Molecule Inhibitors of Human Galactokinase  
Klaas J. Wierenga, MD  
University of Miami

5:30 PM—6:00 PM  Presentations from the 2006 Emmanuel Shapira Award Winners

5:30 PM—5:45 PM  Metabolic Control During Exercise With and Without Medium-Chain Triglyceride (MCT) in Children with Long-Chain 3-Hydroxyacyl-CoA Dehydrogenase (LCHAD) or Trifunctional Protein (TFP) Deficiency
Melanie Gillingham, PhD, RD
Oregon Health & Science University

5:45 PM-6:00 PM Propionyl-CoA and Adenosylcobalamin Metabolism in C. elegans: Evidence for a Role of Methylmalonyl-CoA Epimerase in Intermediary Metabolism
Randy Chandler, MB
NIH, National Human Genome Research Institute

Evening free in Nashville

Tuesday, March 27, 2007

7:00 AM–8:00 AM Breakfast—Renaissance Nashville Hotel—Music City Ballroom—Level Two

8:00 AM–11:30 AM Session III Modifiers of Metabolic Pathways—NCC Ballrooms 204-206—Level Two
Moderator: Katrina Dipple, MD, PhD
David Geffen School of Medicine at UCLA

8:00 AM–8:45 AM Modifiers of Inborn Errors of Metabolism: A Systems Biology Approach
Edward C. McCabe, MD, PhD
Mattel Children’s Hospital at UCLA

8:45 AM–9:30 AM Carrier Status as a Contributor to Complex Disorders
Philip A. Wood, DVM, PhD
University of Alabama at Birmingham

9:30 AM–10:15 AM Multifunctional Regulators of Mitochondrial Iron Balance
Grazia Isaya, MD
Mayo Clinic

10:15 AM–10:45 AM Break—NCC East Exhibit Hall—Level One

10:45 AM–11:30 AM Modifiers of Electron Transport: Coenzyme Q10 Deficiency
Michio Hirano, MD
Columbia University Medical Center

11:45 AM–1:00 PM SIMD Board Meeting—Jazz Room, 4th floor, Renaissance Nashville Hotel

12:30 PM–2:30 PM Organization Meeting for the North America Metabolic Academy—INVITATION ONLY

2:30 PM–3:30 PM 2009 International Meeting—Organizing Committee Meeting
Jazz Room, 4th floor, Renaissance Nashville Hotel

Afternoon Free to Enjoy Nashville

1:00 PM–5:00 PM Posters on display in Convention Center—NCC East Exhibit Hall—Level One

4:00 PM–6:00 PM Posters Attended by Authors—NCC East Exhibit Hall—Level One
Wine and cheese served

6:00 PM–7:00 PM Dinner—(Optional: must be pre-paid)
Renaissance Nashville Hotel—Music City Ballroom—Level Two

7:15 PM–7:45 PM Donough O’Brien Presidential Address—NCC Ballrooms 204-206—Level Two
Jerry Vockley, MD, PhD
University of Pittsburgh

7:45 PM–9:00 PM SIMD Business Meeting and Award Presentations—NCC Ballrooms 204–206
Emmanuel Shapira SIMD Award (First Author of Best Publication in MGM)
Neil Buist Award (Best Oral Presentation by a Trainee)
Wednesday, March 28, 2007

7:00 AM–8:00 AM  
*Breakfast—Renaissance Nashville Hotel—Music City Ballroom—Level Two*

8:00 AM–12:00 Noon  
*NCC Ballrooms 204-206—Level Two*

**Session IV. Treatment of Inborn Errors of Metabolism in the Expanded Newborn Screening Age**

**Moderator:** John Phillips, MD  
Vanderbilt University School of Medicine

8:00 AM–8:45 AM  
**Babies and VLCAD Newborn Screening. What Does Genotype Mean?**

Arnold Strauss, MD  
Vanderbilt University School of Medicine

8:45 AM–9:30 AM  
**Survival of Patients with Urea Cycle Disorders Following Treatment of Hyperammonemia with Intravenous Sodium Phenylacetate and Sodium Benzoate**

Gregory M. Enns, MD  
Stanford University

9:30 AM–10:15 AM  
**An Update on the BioMarin PKU Clinical Development Program**

Alexander Dorenbaum, MD  
BioMarin Pharmaceutical, Inc.

10:15 AM–10:45 AM  
*Coffee Break—NCC East Exhibit Hall—Level One*

10:45 AM–12:00 Noon  
**Ask the Experts: Challenge the Panel with Unusual and Unknown Cases.**  
(Attendees may submit candidate cases in advance or at the meeting)

**Panel Experts:**

Barbara Burton, MD, Children’s Memorial Hospital, Chicago  
Annette Feigenbaum, MD, The Hospital for Sick Children, Toronto  
Stephen G. Kahler, MD, Arkansas Children’s Hospital, Little Rock  
Johan Van Hove, MD, PhD, The Children’s Hospital, Denver

*Adjourn*