



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

Katherine H. Kim, MS, Children's Memorial Hospital, Chicago, IL
Piero Rinaldo, MD, PhD, Mayo Clinic, Rochester, MN
Rani Singh, RD, Emory University, Atlanta, GA

Cases presented by:

Carol L. Greene, MD, University of Maryland (bMCC)
Annette Feigenbaum, MD, The Hospital for Sick Children, Toronto (tyrosinemia)
Nicola Longo, MD, PhD, University of Utah, Salt Lake City, UT (MMA, MCADD)
Rani Singh, RD, Emory University (galactosemia)

7:00 PM–10:00 PM *Welcome Reception—Renaissance Nashville Hotel—East Grand Ballroom Lobby Level*
Sponsored in part by Ucylyd Pharma

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 Mucopolidosis 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 4 α -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*