Program and Abstracts for the SIMD Annual Meeting

Asilomar Conference Center
Pacific Grove, California

Sunday, March 2, 2008

1:00 PM – 6:00 PM  Registration – Asilomar Conference Center – Phoebe A Hearst Hall
3:00 PM – 9:00 PM  Exhibit and poster set up – Fred Farr Forum and Kiln
3:00 PM – 6:00 PM  Asilomar Room Check-in – Phoebe A Hearst Hall
3:30 PM – 6:00 PM  Welcome Dinner – Crocker Dining Hall
7:30 PM – 10:00 PM  Opening Reception – Merrill Hall (Sponsored in part by Actelion Pharmaceuticals)

Monday, March 3, 2008

7:00 AM – 8:00 AM  Breakfast – Crocker Dining Hall
8:00 AM – 12:00 PM  Registration – Merrill Hall

Scientific Session 1: Session 1: Advances in Diagnosis, Management and Monitoring in Inborn Errors of Metabolism – Merrill Hall

8:00 AM – 9:50 AM  Moderator: Mendel Tuchman, M.D.
Children’s National Medical Center, Washington, DC
8:00 AM – 8:40 AM  The Promise of siRNA therapy
Jon Wolff, M.D.
Waisman Center, Madison, WI
8:40 AM – 9:15 AM  New Frontiers in Neuroimaging – Applications to Inborn Errors of Metabolism
Andrea Gropman, M.D.
Children’s National Medical Center, Washington, DC
9:15 AM – 9:50 AM  Lending a Helping Hand: Chaperones and Lysosomal Storage Disease
Don Mahuran, Ph.D.
The Hospital for Sick Children, Toronto, Ontario, Canada

Scientific Session 2: Session 2: Controversies in Protein Requirements in IEM: What We Know, and What We Still Need To Learn - Merrill Hall

10:15 AM – 12:00 PM  Moderator: Barbara Marriage, Ph.D., R.D.
Ross Metabolics/Abbott Laboratories, Columbus, OH
10:15 AM – 10:50 AM  Recent advances in the determination of protein and amino acid requirements in humans - with a focus in inborn errors of amino acid metabolism
Paul Pencharz, M.B., Ch.B., Ph.D.
The Hospital for Sick Children, Toronto, Ontario, Canada
10:50 AM – 11:25 AM  Protein Requirements in Patients with IEM: What’s Needed to Support Optimal Growth and Clinical Outcome?

Rani Singh, Ph.D., R.D.
Emory University, Atlanta, GA

11:25 AM – 12:00 PM  Protein: Maximum Tolerated or Minimum Required – Are they Mutually Exclusive?
Panel Experts:
Paul Pencharz, M.B., B.Ch.B., Ph.D., William Nyhan, M.D., Rani Singh, Ph.D., R.D. and Jean-Marie Saudubray, M.D.

12:00 PM – 1:00 PM  Lunch – Crocker Dining Hall

Scientific Session 3:

1:30 PM – 5:00 PM  Session 3: Contributed Papers
Presentations from Travel Award Winners and Ucyclyd Fellows – Merrill Hall

1:30 PM – 1:45 PM  The mitochondrial G13513A mutation frequency in a Leigh like disease cohort: clinical, biochemical and molecular features of six new patients.
Ariel Brautbar, M.D.
Baylor College of Medicine, Houston, TX

1:45 PM – 2:00 PM  Outcomes Among Newborns with Abnormal C5-OH on Expanded Newborn Screening in California.
Kristina Cusmano-Ozog, M.D.
Stanford University, Stanford, CA

2:00 PM – 2:15 PM  Regulation of Cellular Metabolism by Manipulating Signal Transduction Pathways: Akt and e-Myc Promote an Anabolic Phenotype Favoring Cell Growth
Ralph J. DeBerardinis, M.D., Ph.D.
Children’s Hospital of Philadelphia, Philadelphia, PA

2:15 PM – 2:30 PM  Mutations in the SC4MOL gene cause autosomal recessive psoriasiform dermatitis, revealing a novel mechanism for the pathogenesis of psoriasis
Miao He, Ph.D.
Children’s Hospital of Pittsburgh, Pittsburgh, PA
Travel award winner and Ucyclyd Fellow

2:30 PM – 2:45 PM  In Vitro Effect of Miglustat on Phosphotransferase Deficient I-Cells
Brianne Howarth
McMaster University, Hamilton, Ontario, CANADA

2:45 PM – 3:00 PM  Detection of Remethylation Disorders in Dried Blood Spots by Combined MS/MS and LC/MS/MS Methods
James Lim, Ph.D.
Mayo Clinic, Rochester, MN

3:00 PM – 3:30 PM  Coffee Break – visit exhibits – Fred Farr Forum and Kiln

3:30 PM – 3:45 PM  Urinary measures of oxidative stress in Inborn Errors of Metabolism
Peter McGuire, M.S., M.D.
Mount Sinai Medical Center, New York, NY

3:45 PM – 4:00 PM  Quantitation of Tissue Acyl-CoAs in Beta-Oxidation Defects using Tandem Mass Spectrometry
Andrew Palladino, M.D.
The Children’s Hospital of Philadelphia, Philadelphia, PA
Ucyclyd Fellow
4:00 PM – 4:15 PM Targeted disruption of the Neu4 sialidase gene results in mice with lysosomal storage and abnormal ganglioside catabolism.
Volkan Seyrantepe, Ph.D.
University of Montreal, Quebec, Canada

4:15 PM – 4:30 PM Carnitine supplementation induces acylcarnitine production in tissues of very long-chain acyl-CoA dehydrogenase-deficient mice, without replenishing low free carnitine
Ute Spiekerkoetter, M.D.
Heinrich-Heine University, Duesseldorf, Germany

4:30 PM – 4:45 PM Rescuing a Common Mutation in Hermansky-Pudlak Syndrome Type 1 Using Exon Skipping
Lisa Vincent, Ph.D.
NHGRI/NIH, Bethesda, MD

4:45 PM – 5:00 PM Study of the biophysical relationship between mitochondrial fatty acid oxidation and oxidative phosphorylation.
Yudong Wang, Ph.D.
Children's Hospital of Pittsburgh, Pittsburgh, PA

6:00 PM – 7:00 PM Dinner – Crocker Dining Hall

7:00 PM – 8:15 PM Business Meeting of the SIMD
Barbara Burton, M.D., President; Cary Harding, M.D., Treasurer and other SIMD business and discussion – Merrill Hall

Scientific Session 4:

8:15 PM – 10:00 PM With refreshments

Tuesday, March 4, 2008

7:00 AM – 8:00 AM Breakfast – Crocker Dining Hall

Scientific Session 5:

8:00 AM – 11:15 AM

8:00 AM – 11:15 AM

8:00 AM – 8:35 AM Research Collaboration and Consensus Development in Inborn Errors of Metabolism – Public and Private Resources and opportunities.
Rod Howell, M.D.
National Institute of Child Health and Human Development, NIH, Bethesda, MD

8.35 AM – 9:10 AM Setting up Multi-Institutional Network Research in Rare Diseases: The Urea Cycle Consortium
Mendel Tuchman, M.D.
Children's National Medical Center, Washington, DC

9:10 AM – 9:45 AM Building Consensus for Nutrition Therapy in Patients with IEM
Fran Rohr, M.S., R.D. L.D.
Children's Hospital Boston, Boston, MA

9:45 AM – 10:15 AM Coffee Break – visit exhibits – Fred Farr Forum and Kiln

10:20 AM – 10:55 AM Clinical Definition of Cutoff Values for Newborn Screening of Metabolic Disorders: An Evidence-Based, Collaborative Approach
Piero Rinaldo, M.D., Ph.D.
Mayo Clinic College of Medicine, Rochester, MN

11:00 AM – 11:40 PM Donough O'Brien Presidential Address – Merrill Hall
Barbara Burton, M.D.
Children's Memorial Medical Center, Chicago, IL

11:40 AM – 12:00 PM Award Presentations:
Emmanuel Shapira SIMD Award and Oral Presentation
Expression and characterization of mutations in human very long-chain acyl-CoA dehydrogenase using a prokaryotic system.
Eric Goetzman, Ph.D
University of Pittsburgh, Pittsburgh, PA

Neil Buist Award (Best Oral Presentation by a Trainee)
12:00 PM – 1:00 PM Lunch – Crocker Dining Hall
   Afternoon Free to Enjoy the Beach and the Monterey Peninsula
1:00 PM – 6:00 PM Posters and exhibits on display in Fred Farr Forum and Kiln
Scientific Session 6 Session 6: Poster #2 – Even # poster authors available for discussion - and visit exhibits – Fred Farr Forum and Kiln (Sponsored in part by Genzyme Therapeutics)
4:30 PM – 6:00 PM With refreshments
6:00 PM – 7:00 PM Dinner – Crocker Dining Hall
Scientific Session 7 Session 7: Ask the Experts! Challenge the panel with unusual and unknown cases. (Attendees may submit candidate cases in advance or at the meeting) – Merrill Hall
7:15 PM – 9:00 PM Moderator: Eric Crombez, M.D., Shire HGT, Cambridge, MA
   Panel Experts:
      Georgianne Arnold, M.D., University of Rochester, Rochester, NY
      Gregory Enns, M.D., Stanford University, Stanford, CA
      Johan Van Hove, M.D., Ph.D., University of Colorado @ Denver, Denver, CO

Wednesday, March, 5, 2008
7:00 AM – 8:00 AM Breakfast – Crocker Dining Hall
Scientific Session 8: Session 8: Metabolic Liver Disease – Merrill Hall
8:00 AM – 9:45 AM Moderator: Annette Feigenbaum, M.B., Ch.B.
       The Hospital for Sick Children, Toronto, Ontario, Canada
8:00 AM – 8:35 AM Clinical Approach to Liver Metabolic Diseases: An Overview
       Jean-Marie Saudubray, M.D.
       Hopital Necker Enfants Malades, Paris, France
8:35 AM – 9:10 AM Mitochondrial Hepatopathies: Emerging Genotypes and Clinical Phenotypes
       Ron Sokol, M.D.
       University of Colorado Health Sciences Center, Denver, CO
9:10 AM – 9:45 AM Diagnosis and Treatment of Genetic Defects of Bile Acid Synthesis
       Kenneth Setchell, Ph.D.
       Cincinnati Children’s Hospital Medical Center, Cincinnati, OH
9:45 AM – 10:15 AM Coffee Break – visit exhibits – Fred Farr Forum and Kiln
10:15 AM – 12:05 PM Moderator: Carol Greene, M.D.
       University of Maryland School of Medicine, Baltimore, MD
10:15 AM – 10:50 AM When Should Psychiatry Consult the Metabolic Specialist?
       David Mallott, M.D.
       University of Maryland School of Medicine, Baltimore, MD
10:50 AM – 11:30 AM Whole Genome Approaches to Genetic Variance in Human Metabolism: Lessons from Pharmacogenetics
       M. Eileen Dolan, Ph.D.
       University of Chicago, Chicago, IL
11:30 AM – 12:05 PM Carnitine Palmitoyl Transferase 1A Deficiency and Newborn Screening: Implications for Public Health
       David Koeller, M.D.
       Oregon Health & Science University, Portland, OR
       Adjourn
SIMD ABSTRACTS

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The 2007 Emmanuel Shapira SIMD Award

The Emmanuel Shapira SIMD Award was established in 2003 to recognize the best paper in the field of biochemical genetics and metabolism published in Molecular Genetics and Metabolism (MGM) by an SIMD member or member’s trainee. It was named in memory of Emmanuel Shapira, M.D., Ph.D., one of the founders and most ardent supporters of the Society for many years. Dr. Shapira graduated with an M.D. from Hebrew University in Jerusalem and received his Ph.D. in immunochemistry at the Weizmann Institute of Science in Israel. He was a member of the Faculty of the Department of Pediatrics at Northwestern University Feinberg School of Medicine in Chicago, and later became Professor of Pediatrics and Pathology and Director of the Hayward Genetics Center at Tulane University in New Orleans.

Dr. Shapira’s clinical and research interests were focused on inborn errors of metabolism. He made numerous contributions to the field of biochemical genetics and was a dedicated physician to his patients and a supportive and compassionate teacher to his students. His consistent participation in the annual meetings of the Society contributed both in knowledge and in spirit by making the meetings both scientifically stimulating and enjoyable. This award, that bears his name and the name of the Society that he cherished, is intended to recognize high-quality work in the field that he so loved. A $1,000 prize is awarded annually to the first author of the winning paper chosen by a committee that includes several members of the SIMD and the Editor-in-Chief of MGM.

This year the winner of the Emmanuel Shapira award is Eric Goetzman, Ph.D. for his article entitled “Expression and characterization of mutations in human very long-chain acyl-CoA dehydrogenase using a prokaryotic system”. E.S. Goetzman, Y. Wang, M. He, A.W. Mohsen, B.K. Ninness & J. Vockley: Mol. Gen. & Metab. 91(2), June 2007, p. 138–147. Dr. Goetzman is an Assistant Professor in the Department of Pediatrics at the University of Pittsburgh, Children’s Hospital of Pittsburgh. He will give a brief oral presentation Tuesday, March 4th, 2008 at the Annual Meeting of the SIMD in Asilomar.

Past Winners of the Emmanuel Shapira Award:
2003 Elena Tartaglini, Ph.D.
   Judith C. Fleming, Ph.D.
2004 Gerard T. Berry, M.D.
2006 Randy Chandler, M.B.
   Melanie Gillingham, Ph.D., R.D.

The 2008 Neil Buist Award

The Neil Buist Award was established in 2004 in honor of Dr. Neil Buist, a former President of the SIMD, who served continuously for 26 years on the Board of Directors of the Society until his retirement in 2003. It is awarded annually to the trainee who gives the most outstanding oral presentation at the annual meeting. The winner is selected by a committee of SIMD members during the course of the meeting. A plaque honoring the awardee will be presented this year during the meeting.

Past winners are:
2004 Lina S. Correa-Cerro, M.D., Ph.D.
2005 Amanda Helip-Wooley, Ph.D.
2007 Miao He, Ph.D.
2008 TRAVEL AWARD WINNERS

The winners of the 2008 Travel awards are:

Ariel Brautbar, M.D.
Kristina Cusmano-Ozog, M.D.
Ralph DeBerardinis, M.D., Ph.D.
Shweta Dhar, M.D.
Marni Falk, M.D.
Miao He, Ph.D.
Brianne Howarth
James Lim, Ph.D.
Peter McGuire, M.S., M.D.
Sabrina Mitchell
Volkan Seyrantepe, Ph.D.
Ute Spiekerkoetter, M.D.
Joseph Thakuria, M.D.
Lisa Vincent, Ph.D.
Yudong Wang, Ph.D.

Each recipient received a travel award in the amount of $1,000. Eleven winners of the 2008 Travel awards will give a brief oral presentation Monday afternoon, March 3, 2008 at the SIMD Annual meeting. Four others will present a poster at the SIMD Annual meeting. All have submitted abstracts which are printed in this issue.

Trainee travel awards were supported by a grant from the National Institute of Child Health and Human Development and the Office of Rare Diseases.