

SOCIETY FOR INHERITED METABOLIC DISORDERS

Annual Meeting: March 9 - 12, 2014

Asilomar Conference Center, Pacific Grove, California

Sunday, March 9, 2014

1:00 - 6:00 PM	Meeting Registration - Merrill Hall Poster board and vendor set up - Pavilion
3:00 - 8:00 PM	Asilomar check in - Phoebe A. Hearst Social Hall
3:00 - 6:00 PM	SIMD Board meeting - Surf and Sand (by invitation)
6:00 PM - 7:30 PM	Dinner - Crocker Dining Hall
7:30 - 10:00 PM	Welcome reception - Merrill Hall

Monday, March 10, 2014

7:00 AM - 8:00 AM	Breakfast - Crocker Dining hall
8:00 AM - 5:30 PM	Registration - Merrill Hall Poster set up - Pavilion
Scientific Session 1 8:00 AM - 12:15 PM	Metabolic Testing in the Post Human Genome Project Era Moderators: Tina Cowan & Michael Bennett
8:00 - 8:20 AM	<i>Metabolomic Approaches to Metabolic Testing</i> Bruce Barshop, M.D., Ph.D. University of California, San Diego, La Jolla, CA
8:20 - 8:40 AM	<i>Genomic Sequencing in the Metabolic Clinic</i> David Dimmock, M.D. Medical College of Wisconsin, Milwaukee, WI
8:40 - 9:00 AM	<i>Diagnosing CDG's: Beyond Isoelectric Focusing</i> Lynne Wolfe, M.S., C.R.N.P., B.C. NIH/NHGRI, Bethesda, MD
9:00 - 9:40 AM	<i>Human iPSC: Promise for Diagnosis, Modeling, Drug Screening and Cell Based Therapies for IEMs</i> Juan Carlos Izpisua Belmonte, Ph.D. The Salk Institute for Biological Studies, La Jolla, CA
9:40 - 10:30 AM	AM Coffee break - Exhibits open - Pavilion
Scientific Session 2 10:30 AM - 12:00 PM	Purine and Pyrimidine Metabolism Moderators: Carol Greene & Cynthia Freehauf
10:30 - 11:00 AM	<i>The Biochemistry and Pathophysiology of Purine and Pyrimidine Metabolism</i> A.B.P. van Kuilenburg, Ph.D. Academic Medical Center, Amsterdam, The Netherlands
11:00 - 11:20 AM	<i>Lesch Nyhan Disease - An Historical Perspective and Update</i> William Nyhan, MD, PhD University of California, San Diego, La Jolla, CA

11:20 - 11:40 AM	<i>Diagnostic Testing for Purine and Pyrimidine Related Intellectual Disability</i> V. Reid Sutton, M.D. Baylor College of Medicine & Texas Children's Hospital, Houston, TX
11:40 - 12:00 Noon	<i>Purine and Pyrimidine Nucleoside Synthesis, Recycling and Transport Defects</i> Michio Hirano, M.D. Columbia University Medical Center, New York, NY
12:00 - 1:00 PM	Lunch - Crocker Dining Hall
Scientific Session 3 1:00 - 2:30 PM	Award Recipients and Submitted Abstracts Presentations Moderators: Shawn McCandless & Cary Harding
1:00 - 1:15 PM	<i>Functional Characterization and Categorization of Missense Mutations that Cause Methylmalonyl-CoA Mutase Deficiency</i> Patrick Forny, M.D. University Children's Hospital, Zurich, Switzerland
1:15 - 1:30 PM	<i>Validation and Application of a LC-HRMS Platform for Simultaneous Quantification and Biomarker Discovery in the Clinical Laboratory</i> Ilya Gertsman, Ph.D. University of California, San Diego, San Diego, CA
1:30 - 1:45 PM	<i>Administration of AAV Vectors to Pcca-/(A138T) Mice Elicits Long Term Correction of Disease Markers</i> Adam J. Guenzel Mayo Clinic College of Medicine, Rochester, MN
1:45 - 2:00 PM	<i>Adenosine Deaminase 2 (ADA2) Deficiency: A Novel Inborn Error of Purine Metabolism</i> Ben Pode-Shakked, M.D. Safra Children's Hospital, Sheba Medical Center, Tel-Hashomer, Israel
2:00 - 2:15 PM	<i>Moonlighting in Mitochondria: ACAD9 Plays a Dual Role in Energy Metabolism</i> Manuel Schiff, M.D., Ph.D. University of Pittsburgh, Pittsburgh, PA
2:15 - 2:30 PM	<i>Combined Triple Therapy Trial of Pyridoxine, Lysine Restricted Diet and Arginine Supplementation in Four Patients with Pyridoxine-Dependent Epilepsy</i> Andrew Shuen, HBSoc, M.D. Montreal Children's Hospital, McGill University, Montreal, Quebec
2:30 - 2:45 PM	Presentation from the 2013 Emmanuel Shapira Award Winner <i>Pathogenesis of growth failure and partial reversal with gene therapy in murine and canine glycogen storage disease type 1</i> Elizabeth Drake Brooks, M.S., D.V.M. Duke University, Durham, NC

2:45 - 3:00 PM	SIMD/Hyperion Fellow Research Presentation <i>Drug Screening Using a Drosophila Model of Mitochondrial Complex I Deficiency and Oxidative Stress</i> Zhihong Li, MD, PhD, Postdoctoral Associate Baylor College of Medicine
3:00 - 4:00 PM	PM coffee break - Pavilion
Scientific Session 4 4:00 - 5:30 PM	Submitted Abstracts Presentations Moderators: Shawn McCandless & Nicola Longo
4:00 - 4:15 PM	<i>Neuroprotection from Hyperammonemia: a Small Molecule Screen</i> Ljubica Caldovic, Ph.D. Children's National Medical Center, Washington, DC
4:15 - 4:30 PM	<i>Mutations in NGLY1 Cause an Inherited Disorder of the Endoplasmic Reticulum-Associated Degradation (ERAD) Pathway</i> Gregory Enns, M.D. Stanford University, Stanford, CA
4:30 - 4:45 PM	<i>Second-Tier DNA Confirmation of Newborn Metabolic and Hearing Loss Screening Using Targeted Next Generation Sequencing</i> Ed Naylor, Ph.D., M.P.H. Parabase Genomics, Boston, MA
4:45 - 5:00 PM	<i>Clinical Outcome in Children with UCD after Liver Cell Therapy in Comparison to a Control Population</i> Thomas Opladen, M.D. University Children's Hospital, Heidelberg, Germany
5:00 - 5:15 PM	<i>Psychometabolomics: Assessment of Treatment-Refractory Depression</i> Lisa Pan, M.D. University of Pittsburgh, Pittsburgh, PA
5:15 - 5:30 PM	<i>Outcome in nonketotic hyperglycinemia</i> Johan Van Hove, M.D., Ph.D. University of Colorado, Aurora, CO
6:00 - 7:00 PM	Dinner - Crocker Dining Hall
Scientific Session 5 7:15 - 9:30 PM	Posters Attended by Authors and Exhibits open - Pavilion Posters open to all. Wine and dessert served
7:30 - 8:25 PM	Travel Award Poster Presentations (5 minute presentation at each poster from travel award winners attended by Buist Committee)
7:30 - 7:35 PM Poster Board #12	<i>Rapid Point of Care Detection of Hyperammonemia in Whole Blood</i> Omar Ayyub, B.S. & Ph.D. candidate, Bioengineering University of Maryland, College Park, MD

7:40 - 7:45 PM Poster Board #28	<i>Quantification of Urine Keratan Sulfate Levels in Mucopolidosis II and III Patients by UPLC-MS/MS</i> Katarzyna (Kasia) Ellsworth, Ph.D. Greenwood Genetics Center, Greenwood, SC
7:50 - 7:55 PM Poster Board #35	<i>Diagnosis of Primary Hyperoxaluria Type III, A Novel Disorder of Hydroxyproline Metabolism, by GC-MS Analysis of Urine</i> Linda Hasadsri M.D., Ph.D. Mayo Clinic, Rochester, MN
8:00 - 8:05 PM Poster Board #52	<i>Unraveling a Neurometabolic Hereditary Spastic Paraplegia: B4GALNT1 Deficiency as New Inborn Error of Metabolism Affecting Glycosphingolipid Biosynthesis</i> Charles Marques Lourenco, M.D. Clinics Hospital of Ribeirao Preto - School of Medicine of Ribeirao Preto, University of Sao Paulo, Sao Paulo, Brazil
8:10 - 8:15 PM Poster Board #61	<i>Oxidative Stress Induces Mitochondrial Dysfunction in Autism Cell Lines</i> Shannon Rose, Ph.D. Arkansas Children's Hospital Research Institute, Little Rock, AR
8:20 - 8:25 PM Poster Board #79	<i>Holding Parenteral Nutrition for 3 Hours Before Newborn Screening Markedly Reduces False Positive Amino Acid Results</i> Thipwimol Tim-aroon, M.D. University Hospitals Case Medical Center, Cleveland, OH

Tuesday, March 11, 2014

7:00 - 8:00 AM	Breakfast - Crocker Dining Hall
8:00 AM -12:00 PM	Registration - Merrill Hall
Scientific Session 6 8:00 -10:10 AM	50 Years of Newborn Screening: Where Are We Now and How Did We Get Here? Moderators: Georgianne Arnold & Melanie Gillingham
8:00 - 8:40 AM	<i>NBS - The First 50 Years</i> Harvey Levy, M.D. Children's Hospital Boston, Boston, MA
8:40 - 9:10 AM	<i>The Realpolitik of Newborn Screening</i> Rodney Howell, MD University of Miami, Miller School of Medicine, Miami, FL
9:10 - 9:40 AM	<i>Dietary treatment for inherited metabolic disorders detected through NBS: past, present and future</i> Dianne Frazier, PhD, MPH University of North Carolina at Chapel Hill, Chapel Hill, NC
9:40 - 10:10 AM	<i>Do Wilson and Jungner Still Matter?</i> Lainie Ross, MD, PhD University of Chicago, Chicago, IL
10:10 -10:40 AM	AM Coffee break - Exhibits open - Pavilion

Scientific Session 7 10:40 AM -12:00 PM	50 Years of Newborn Screening: Where Are We Going? Moderators: Sandy van Calcar & Dietrich Matern
10:40 - 11:00 AM	<i>Collecting the data - NBSTRN, REDCap and Regional Databases</i> Sue Berry, MD University of Minnesota, Minneapolis, MN
11:00 - 11:20 AM	<i>NBS: Adding Geno(me)typing to Pheno(me)typing</i> Bob Nussbaum, M.D. Institute for Human Genetics, UC San Francisco, San Francisco, CA
11:20 - 11:40 AM	<i>Role of the SIMD in Guiding Future Directions for NBS</i> Neil Buist, MB, ChB (founding member, SIMD) Oregon Health & Science University, Portland, OR
11:40 AM - 12:20 PM	<i>The Robert Guthrie Memorial Lecture: How Newborn Screening Moves Forward</i> David Millington, PhD Duke University School of Medicine, Durham, NC
12:30 -1:30 PM	Lunch - Crocker Dining Hall
12:30 -1:30 PM	How to Get Published. Author Workshop. Presented by Paige Shaklee, Publisher, <i>Genomics</i> - Crocker Dining Hall (see info)
1:30 - 6:00 PM	Free afternoon in Asilomar
1:00 - 6:00 PM	Posters on display - Pavilion
4:00 - 6:00 PM	NAMA Reception - by invitation only - Surf and Sand
6:00 - 7:00 PM	Dinner - Crocker Dining Hall
7:30 - 8:00 PM	Presidential Address - Merrill Hall <i>Membrane Transport and Metabolic Disorders</i> Nicola Longo, M.D., Ph.D., SIMD President University of Utah, Salt Lake City, UT
8:00 - 9:30 PM	SIMD Business Meeting and Award Presentations Emmanuel Shapira SIMD Award (First Author of Best Publication in MGM) Neil Buist Award (Best Oral Presentation by a Trainee)
Wednesday, March 12, 2014	
7:00 - 8:00 AM	Breakfast - Crocker Dining Hall
Scientific Session 8 8:00 -10:00 AM	The Brain in IEMs Moderators: Katrina Dipple & Fernando Scaglia

8:00 - 8:40 AM	<i>Inborn Errors of Creatine and Serine Synthesis</i> Sylvia Stöckler, MD British Columbia Children's Hospital, Vancouver, BC
8:40 - 9:05 AM	<i>GABA and Mitophagy: Novel Pathomechanisms in Disorders of GABA Metabolism</i> Mike Gibson, PhD Washington State University, College of Pharmacy, Spokane, WA
9:05 - 9:30 AM	<i>D-2-Hydroxyglutaric Aciduria</i> Gajja Salomons, PhD VU University Medical Center, Amsterdam, The Netherlands
9:30 - 10:00 AM	<i>Diagnosis and Discovery of Treatable Inborn Errors of Metabolism causing Intellectual Disability</i> Clara van Karnebeek, MD, PhD, University of British Columbia, Vancouver, BC
10:00 - 10:30 AM	AM Coffee Break - Pavilion
Scientific Session 9	The Brain in IEMs (continued)
10:30 AM - 11:45 AM	Moderators: Marshall Summar & Nancy Braverman
10:30 AM - 10:55 AM	<i>Role of Inflammation in Brain Injury During Metabolic Decompensation</i> George Diaz, M.D. Mount Sinai School of Medicine, New York, NY
10:55 AM- 11:20 AM	<i>Preventing Brain Injury in Glutaric Aciduria Type 1</i> Stefan Kölker, MD University Children's Hospital, Heidelberg, Germany
11:20 AM - 11:45 AM	<i>Neuroprotection During Acute Metabolic Decompensation</i> Uta Lichter, M.D., Ph.D. Children's National Medical Center, Washington, DC
11:45-12:00	Closing Comments