# PROGRAM: SOCIETY FOR INHERITED METABOLIC DISORDERS

## Annual Meeting: March 28 - 31, 2015
## Grand America Hotel, Salt Lake City, Utah

| | |
|---|---|
| **Friday, March 27, 2015** | |
| 4:00 PM – 7:00 PM | Meeting Registration – Grand Ballroom Registration Desk |
| **Saturday, March 28, 2015** | |
| 8:30 AM - 6:00 PM | Meeting Registration – Grand Ballroom Registration Desk |
| 1:00 PM– 5:00 PM | Poster board and vendor set up – Imperial Ballroom |
| **Scientific Session 1**<br>10:30 AM – 12:00 PM | Joint Session with ACMG – Salt Palace Convention Center<br>**Could it be metabolic? Practical Approaches to Common Presentations**<br>Moderators: Georgianne Arnold and Shawn E. McCandless |
| 10:30 AM – 11:00 AM | *Diagnostic approach to metabolic hepatomegaly*<br>Jean Marie Saudubray, M.D.<br>Senior metabolic consultant neurometabolic unit La Pitié Salpêtrière Hospital (APHP) Paris, France |
| 11:00 AM – 11:30 AM | *Floppy Babies: How to think your way through the evaluation*<br>Mark Korson, M.D.<br>The Institute for Metabolic Education, Pittsburgh, PA/Boston, MA |
| 11:30 AM – 12:00 PM | *Hypoglycemia: When is it Metabolic?*<br>Georgianne Arnold, M.D., University of Pittsburgh, Pittsburgh, PA |
| 12:00 PM – 1:00 PM | Lunch (on your own) |
| **Scientific Session 2**<br>1:00 PM - 3:00 PM | Joint Session with ACMG – Salt Palace Convention Center<br>**Diagnosis of Metabolic Disease in the 21st Century**<br>Moderators: Dietrich Matern and Georgianne Arnold |
| 1:00 PM – 1:40 PM | *The NIH Undiagnosed Disease Program: Shedding light on the future of diagnosis of metabolic disease*<br>William A. Gahl, M.D., Ph.D.<br>National Human Genome Research Institute/NIH, Bethesda, MD |
| 1:40 PM – 2:20 PM | *A web-based software for covariate-adjusted pattern recognition and integrated reporting of IEM biochemical phenotypes*<br>Piero Rinaldo, M.D., Ph.D.<br>Mayo Clinic, Rochester, MN |
| 2:20 PM – 3:00 PM | *Treatable Neurometabolic Disorders*<br>Clara van Karnebeek, M.D., Ph.D., FCCMG<br>BC Children's Hospital, University of British Columbia, Vancouver, BC |
| 3:00 PM - 6:00 PM | SIMD Board meeting – Embassy |
| 4:00 PM – 6:00 PM | NAMA Board meeting - Ambassador |
| 7:00 PM – 10:00 PM | Opening Reception – Grand Salon |

| | |
|---|---|
| **Sunday, March 29, 2015** | |
| 7:00 AM – 8:00 AM | Breakfast – Grand Ballroom B |
| 7:30 AM – 5:00 PM | Meeting Registration - Grand Ballroom Registration Desk |
| **Scientific Session 3**<br>8:00 AM –10:00 AM | **Biomedical Engineering/Therapies: Bench to Bedside**<br>**Grand Ballroom A & D**<br>Moderators: Dwight Koeberl and Sue Berry |

| | |
|---|---|
| 8:00 AM – 8:35 AM | *Bioengineered human muscle for physiological studies and disease modeling*<br>Nenad Bursac, Ph.D.<br>Duke University, Durham, NC |
| 8:35 AM – 9:10 AM | *Genome Engineering to Correct and Model Genetic Diseases*<br>Charles A. Gersbach, Ph.D.<br>Duke University Medical Center, Durham, NC |
| 9:10 AM – 9:45 AM | *Mitochondrial Replacement Therapy*<br>Shouhkrat Mitalipov, Ph.D.<br>Oregon National Primate Research Center, Oregon Health & Science University, Portland, OR |
| 9:45 AM – 10:15 AM | AM Coffee break - Posters and Exhibits open – Imperial Ballroom |
| 10:15 AM – 10:50 AM | *Chimeric animals as models of rare disease research*<br>Markus Grompe, M.D.<br>Oregon Health & Science University, Portland, OR |
| 10:50 AM – 11:25 AM | *Liver and liver/kidney transplantation in Organic Acidemias*<br>Hilary Vernon, M.D., Ph.D.<br>McKusick-Nathans Institute of Genetic Medicine, John Hopkins University Baltimore, MD |
| **Scientific Session 4**<br>11:25 AM – 12:10 PM | **New Ideas: The Gut Microbiome as a Metabolic Organ**<br>**Grand Ballroom A & D**<br>Moderators: Shawn E. McCandless |
| 11:25 AM – 12:10 PM | *Gut microbiome metabolism of choline and L-carnitine and cardiovascular risk: Implications for metabolic disorders*<br>W.H.Wilson Tang, M.D., F.A.C.C., F.A.H.A.<br>Cleveland Clinic, Lerner College of Medicine at CWRU, Cleveland, OH |
| 12:10 PM –1:00 PM | Lunch – Grand Ballroom B |
| **Scientific Session 5**<br>1:00 PM – 3:00 PM | **Oral Presentations from Travel Award Winners**<br>**Grand Ballroom A & D**<br>Moderators: Melanie Gillingham and Carol Greene |
| 1:00 PM – 1:15 PM | *Long-term Complications of Glycogen Storage Disease Type IA in the dog model treated with gene replacement therapy*<br>Elizabeth D. Brooks, MS., DVM<br>Duke University Medical Center, Durham, NC |
| 1:15 PM – 1:30 PM | *N-Acetylmannosamine (ManNAc) is Safe and Increased Sialic Acid Production in GNE Myopathy Subjects*<br>Brad Class, B.A.<br>National Center for Advancing Translational Sciences (NCATS), NIH Bethesda, MD |
| 1:30 PM – 1:45 PM | *Human IPS cell models of cholesterol synthesis disorders reveal WNT signaling defects underlie neurological dysfunction*<br>Kevin Francis, Ph.D.<br>NIH/NICHD, Bethesda, MD |
| 1:45 PM – 2:00 PM | *Clarifying the phenotype of NGLY1 deficiency, the first congenital disorder of deglycosylation*<br>Christina T. Lam, M.D.<br>NHGRI/NIH, Bethesda, MD |
| 2:00 PM – 2:15 PM | *Zinc finger nuclease-induced targeted integration of a glucose-6- phosphatase gene promotes survival in mice with glycogen storage disease type 1A*<br>Dustin J. Landau, B.S.<br>Duke University Medical Center, Durham, NC |
| 2:15 PM – 2:30 PM | *Successful diagnosis of HIBCH deficiency from exome sequencing and positive retrospective analysis of newborn screening cards in two siblings presenting with Leigh's disease*<br>Ashlee Stiles, Ph.D.<br>Children's Hospital of Orange County, Orange, CA |

| | |
|---|---|
| 2:30 PM – 2:45 PM | **Presentations from the 2014 Emmanuel Shapira Award Winner**<br>*Guanidinoacetate methyltransferase (GAMT) deficiency: Outcomes in 48 individuals and recommendations for diagnosis, treatment and monitoring*<br>Sylvia Stockler-Ipsiroglu, M.D.<br>British Columbia Children's Hospital, Vancouver, BC Canada |
| 2:45 PM - 3:00 PM | **JSIMD Young Investigator award for best presentation at 2014 JSIMD meeting**<br>*GDF15 is a novel biomarker of mitochondrial diseases*<br>Shuichi Yatsuga, M.D., Ph.D.<br>Kurume University School of Medicine, Fukuoka, Japan |
| 3:00 PM – 4:00 PM | PM coffee break - Posters and Exhibits open - Imperial Ballroom |
| **Scientific Session 6**<br>4:00 PM – 5:30 PM | **Platform Presentations --** Selected Abstracts Presentation – new and exciting work of broad general interest to the SIMD membership<br>**Grand Ballroom A & D**<br>Moderators: Bruce Barshop and Cate Walsh Vockley |
| 4:00 PM – 4:15 PM | *The first two years of full population pilot newborn screening for Lysosomal Storage Disorders: the Missouri experience*<br>Andrea M. Atherton, MS.,CGC<br>Children's Mercy Hospital, Kansas City, MO |
| 4:15 PM – 4:30 PM | *ABAT is a novel human mitochondrial DNA depletion syndrome gene linking Gamma-Aminobutyric Acid (GABA) catabolism and mitochondrial nucleoside metabolism*<br>Penelope E. Bonnen, Ph.D. (presenting in place of Arnaud Besse, Ph.D.)<br>Baylor College of Medicine, Houston, TX |
| 4:30 PM – 4:45 PM | *Odd or even? Results from a randomized trial of triheptanoin compared to MCT in patients with long-chain fatty acid oxidation disorders*<br>Melanie Gillingham, Ph.D., RD<br>Oregon Health & Science University, Portland, OR |
| 4:45 PM – 5:00 PM | *Carbonic Anhydrase VA deficiency: an important differential diagnosis of early onset hyperammonemia*<br>Carmen Diez-Fernandez, Ph.D.<br>University Children's Hospital Zurich, Zurich, Switzerland |
| 5:00 PM – 5:15 PM | *Urinary glucose tetrasaccharide concentrations in patients with infantile and late-onset Pompe disease identified by newborn screening*<br>Sarah Young, Ph.D., FACMG<br>Duke University Medical Center, Durham, NC |
| 5:15 PM – 5:30 PM | *NGLY1 deficiency lies in the crosshairs of CDG and mitochondrial diseases*<br>Miao He, Ph.D.<br>University of Pennsylvania, The Children's Hospital of Philadelphia, Philadelphia, PA |
| 5:30 PM - 7:00 PM | Optional Dinner to purchase (ticket required) – Grand Salon |
| **Scientific Session 7** | **Poster Session and Reception– Imperial Ballroom** |
| 7:15 PM – 9:30 PM | **Posters Attended by Authors and Exhibits open**– Posters open to all<br>Wine and dessert served |
| 7:30 PM – 8:45 PM | **Travel Award Poster Presentations** (10 minute presentation at each poster from travel award winner attended by Founders' Award Committee) |
| 7:30 PM – 7:40 PM<br>Poster board #9 | *Development of a rapid point-of-care blood phenylalanine meter for at home and bedside use*<br>Omar Ayyub, Ph.D.<br>Children's National Medical Center, Washington, DC |
| 7:40 PM– 7:50 PM<br>Poster board #30 | *Identification and Characterization of the Biochemical and Physiological Functions of Acyl-CoA Dehydrogenase 10*<br>Kaitlyn Bloom, Ph.D.<br>The Children's Hospital of Philadelphia, Philadelphia, PA |

| | |
|---|---|
| 7:50 PM – 8:00 PM<br>Poster board #50 | *In Vivo OXPHOS Quantitation by Magnetic Resonance Imaging in Metabolic Myopathy*<br>Catherine M. DeBrosse. B.S.<br>University of Pennsylvania, Philadelphia, PA |
| 8:00 PM – 8:10 PM<br>Poster board #19 | *Mutations in MTIF2 cause a novel disorder of mitochondrial translation*<br>Rebecca Ganetzky, M.D.<br>Children's Hospital of Philadelphia, Philadelphia, PA |
| 8:10 PM – 8:20 PM<br>Poster board #64 | *Diagnosing patients with methylmalonic aciduria - comparison of somatic cell and next generation sequencing panel testing*<br>Mihaela Pupavac, Ph.D. Candidate<br>McGill University, Montreal, Quebec |
| 8:20 PM – 8:30 PM<br>Poster board #81 | *GM1 Gangliosidosis Type II: sub-classification of late infantile and juvenile patients based on age at symptom onset*<br>Debra Regier, M.D., Ph.D.<br>NHGRI/NIH, Bethesda, MD |
| 8:30PM – 8:45PM | Committee review |

## Monday, March 30, 2015

| | |
|---|---|
| 7:00 AM – 8:00 AM | Breakfast – Grand Ballroom B |
| 8:00 AM –12:00 PM | Meeting Registration – Grand Ballroom Registration Desk |
| **Scientific Session 8**<br>8:00 AM –10:00 AM | **Membrane Transporter Defects in Humans**<br>**Grand Ballroom A & D**<br>Moderators: Nicola Longo and Laurie Smith |
| 8:00 AM – 8:35 AM | *Families of membrane transporters in humans*<br>Vadivel Ganapathy, Ph.D.<br>Texas Tech University Health Sciences Center, Lubbock, TX |
| 8:35 AM – 9:03 AM | *Lysinuric protein intolerance*<br>Carlo Dionisi-Vici, M.D.<br>Pediatrics Hospital Bambino Gesu', Rome, Italy |
| 9:03 AM – 9:31 AM | *The expanding phenotype caused by riboflavin transporter deficiency*<br>William Rhead, M.D., Ph.D.<br>Medical College of Wisconsin, Milwaukee, WI |
| 9:31 AM – 10:00 AM | *Citrin deficiency and the NADH shuttle*<br>Fernando Scaglia, M.D.<br>Baylor College of Medicine, Houston, TX |
| 10:00 AM – 11:00 AM | AM Coffee break - Posters and Exhibits open – Imperial Ballroom |
| **Scientific Session 9**<br>11:00 AM –12:30 PM | **The Porphyrias Consortium of the NIH Rare Diseases Clinical Research Network (RDCRN): The Inborn Errors of Heme Biosynthesis**<br>**Grand Ballroom A & D**<br>Moderators: Robert Desnick and Marshall Summar |
| 11:00 AM – 11:05 AM | *Introduction on the RDCRN*<br>Robert Desnick, Ph.D., M.D.<br>Icahn School of Medicine at Mount Sinai, New York, NY |
| 11:05 AM – 11:30 AM | *Heme Biosynthesis: Biochemistry, Biology, and Structural Biology*<br>John D. Phillips, Ph.D.<br>University of Utah Health Science Center, Salt Lake City, UT |
| 11:30 AM – 12:00 PM | *The Porphyrias: Classification and Manifestations*<br>Karl E. Anderson, M.D., FACP<br>University of Texas Medical Branch/UTMB Health, Galveston, TX |
| 12:00 PM – 12:30 PM | *Diagnosis and Treatment of the Porphyrias*<br>Robert Desnick, Ph.D., M.D.<br>Icahn School of Medicine at Mount Sinai, New York, NY |
| 12:30 PM – 7:30 PM | Lunch and dinner on your own – Free Afternoon |
| 1:00 PM - 6:00 PM | Posters on display |

| | |
|---|---|
| 5:30 PM – 7:30 PM | **NAMA Reception (by invitation only)**<br>Murano |
| 7:30 PM –8:00 PM | **Past Presidential Address - Grand Ballroom A & D**<br>*Presidential Address: A quarter century later*<br>C. Ron Scott, M.D.<br>University of Washington, Seattle, WA |
| 8:00 PM – 9:30 PM | **SIMD Business Meeting and Award Presentations**<br>Emmanuel Shapira SIMD Award (First Author of Best Publication in MGM)<br>Founders' Award (Best Oral Presentation by a Trainee) |

## Tuesday, March 31, 2015

| | |
|---|---|
| 7:00 AM – 8:00 AM | Breakfast – Grand Ballroom B |
| 7:30 AM – 10:00 AM | Meeting Registration - Grand Ballroom Registration Desk |
| **Scientific Session 10**<br>8:00 AM –10:00 AM | **Inborn Errors of Metabolism and Cancer - Grand Ballroom A & D**<br>Moderators: Fernando Scaglia and Lynne Wolfe |
| 8:00 AM – 8:30 AM | *The role of succinate dehydrogenase and fumarate hydratase in tumorigenesis*<br>Maya Lodish, M.D., MHSc<br>National Institute of Child Health and Human Development/NIH, Bethesda, MD |
| 8:30 AM – 9:00 AM | *The incidence of tumors in lysosomal storage disorders*<br>Greg Pastores, M.D.<br>Yale University School of Medicine, New Haven, CT |
| 9:00 AM – 9:30 AM | *The association of Mitochondrial Hepatopathies and Childhood Liver Cancer*<br>Ron Sokol, M.D.<br>University of Colorado School of Medicine, Aurora, CO |
| 9:30 AM – 10:00 AM | *2-hydroxyglutarate and metabolic reprogramming in cancer*<br>Ralph DeBerardinis, M.D., Ph.D.<br>University of Texas Southwestern, Dallas, TX |
| 10:00 AM -10:30 AM | AM Coffee Break –Posters and Exhibits open - Imperial Ballroom |
| **Scientific Session 11**<br>10:30 AM – 11:45 AM | **Controversies in Newborn Screening for Inborn Errors of Metabolism - Grand Ballroom A & D**<br>Moderators: Katrina Dipple and Marzia Pasquali |
| 10:30 AM – 10:50 AM | *SCAD Deficiency: Differential Diagnosis and Relevance to Human health*<br>William Rhead, M.D., Ph.D.<br>Medical College of Wisconsin, Milwaukee, WI |
| 10:50 AM – 11:10AM | *3-Methylcrotonyl-CoA Carboxylase-deficiency: To screen or not to screen?*<br>Prof. Matthias R. Baumgartner, M.D.<br>University Children's Hospital, Zurich, Switzerland |
| 11:10 AM – 11:45 AM | *Short chain Acyl-CoA Dehydrogenase(SCAD) and 3-methyl-Crotonyl-CoA Carboxylase (3MCC) deficiencies and newborn screening*<br>Marzia Pasquali, Ph.D., FACMG<br>University of Utah School of Medicine, Salt Lake City, UT |
| 11:45 AM -12:00PM | **Closing Remarks** |