SIMD 2022

April 10-13, 2022
Rosen Shingle Creek
Orlando, FL

Sunday, April 10, 2022	
1:00 PM - 6:00 PM	Meeting Registration - Panzacola Registration – Level 1
1:00 PM - 6:00 PM	Poster board and vendor set up - Panzacola F – Level 1
3:00 PM - 6:00 PM	SIMD Board meeting - Suwannee 11 – Level 2
3:00 PM - 6:00 PM	NAMA Board meeting - Suwannee 12 – Level 2
7:00 PM - 10:00 PM	Opening Reception - Gatlin Terrace – Level 1
Monday, April 11, 2022	
7:00 AM - 8:00 AM	Breakfast - Gatline Terrace – Level 1 New to SIMD, new to Rare Disease, or want to meet for breakfast? Join us at the tables labeled "New to SIMD"
7:30 AM – 5:00 PM	Meeting registration - Panzacola Registration – Level 1
Scientific Session 1 (8:00 AM -10:00 AM)	The Continuum of Hope: a session on N-of-1 treatments for inborn errors of metabolism Panzacola G – Level 1 Moderators: Debra Regier, M.D., Ph.D.
8:00 AM - 8:15 AM	Introduction and Welcome Debra Regier, M.D. Ph.D. 2022 Program Chairman Children's National Hospital, Washington DC
8:15 AM - 9:00 AM	Individualized antisense oligonucleotide therapies and the N-of-1 Collaborative: opportunities and challenges Timothy Yu, M.D., Ph.D. Associate Professor in Pediatrics, Division of Genetics and Genomics, Harvard Medical School, Boston, MA
9:00 AM - 9:15 AM	The Continuum of Hope: Navigating the n-of-1 Ethical Landscape Lynn Bush, Ph.D., M.S., M.A. Faculty and Research Associate , Boston Children's Hospital, Lecturer on Pediatrics and Faculty Center for Bioethics, Harvard Medical School, Boston, MA
9:15 AM - 9:30 AM	Guiding a family through development of a custom ASO therapy Austin Larson, M.D. Assistant Professor Pediatrics-Clinical Genetics and Metabolism, Children's Hospital Colorado, Aurora, CO
9:30 AM - 10:00 AM	Panel discussion
10:00 AM-11:00 AM	AM Coffee Break-Posters and Exhibits open – Panzacola F – Level 1
Scientific Session 2 (11:00 AM - 12:00 PM)	Presidential address - Panzacola G – Level 1 An overview of Galactosemia Gerard Berry, M.D. SIMD President Professor of Pediatrics, Harvard Medical School, Boston Children's Hospital, Boston, MA Moderators: Georgianne Arnold, MD, Laurie Bernstein, M.S., R.D.
12:00 PM -1:00 PM	Box Lunch - Panzacola East Foyer – Level 1

Scientific Session 3 (1:00 PM - 3:00 PM)	Oral presentations from Travel Award winners and the 2021 Shapira Award Winner Panzacola G – Level 1 Moderators: Curtis Coughlin, II, Ph.D., MS, MBE and Lindsay Burrage, M.D., Ph.D.
1:00 PM - 1:15 PM	Pharmacology study of AAV vectors for genome editing in GSD 1A infantile mice Benjamin Arnson, Ph.D. Candidate Duke University School of Medicine, Durham, NC
1:15 PM - 1:30 PM	Discovery of plasma N-glycan biomarkers for PMM2-CDG severity Earnest James Paul Daniel, Ph.D. The Children's Hospital of Philadelphia, Philadelphia, PA
1:30 PM - 1:45 PM	Aberrant post-translational modifications characterize the hepatic proteome of methylmalonic acidemia PamelaSara Elbaz Head, Ph.D. National Institute of Health, Bethesda, MD
1:45 PM - 2:00 PM	Multiomics studies in two novel Barth syndrome cell models reveal dysregulation in mitochondrial bioenergetics and quality control: new implications for cellular pathogenesis and therapeutics Olivia Sniezek, B.S. Department of Genetic Medicine, John Hopkins School of Medicine, Baltimore, MD
2:00 PM - 2:15 PM	Mitochondrial-ATP6-associated disease presents with distinct pattern on newborn screening: should it be included as a secondary condition? Christina G. Tise, M.D., Ph.D. Stanford University, Stanford, CA
2:15 PM - 2:30 PM	Cardiolipin profile and mitochondrial bioenergetics alterations in trifunctional protein deficiency Eduardo Vieira Neto, M.D., Ph.D. Genetic and Genomic Medicine, Division of Pediatrics, University of Pittsburgh, Pittsburgh, PA
2:30 PM - 2:45 PM	Presentation from the 2021 Emmanuel Shapira Award Winner - TBD
3:00 PM - 4:00 PM	PM Coffee break – Posters and Exhibits open - Panzacola F – Level 1
Scientific Session 4 (4:00 PM - 5:30 PM)	Selected abstract presentations Panzacola G – Level 1 Moderators: Austin Larson, M.D. and Markey McNutt II, M.D., Ph.D.
4:00 PM - 4:15 PM	Hematopoietic stem cell gene therapy for cystinosis (Phase I/II clinical trial): updated results Bruce Barshop, M.D., Ph.D. Department of Pediatrics, University of California, San Diego, San Diego, CA
4:15 PM - 4:30 PM	Cardiac complications in adults with propionic acidemia Teodoro Jerves, M.D. National Human Research Genome Institute, NIH, Bethesda, MD
4:30 PM - 4:45 PM	C4-OH carnitine: an important marker of ketosis in patients with and without inborn errors of metabolism Maria Laura Duque Lasio, M.D. Department of Pathology, University of Utah, Salt Lake City, UT
4:45 PM - 5:00 PM	Postmortem screening and the impact of newborn screening on mortality rates for MCAD deficiency Freyr Johannsson, Ph.D. Biochemical Genetics Laboratory, Department of Laboratory Medicine and Pathology, Mayo Clinic, Rochester, MN
5:00 PM - 5:15 PM	Neuroimaging findings in inborn errors of metabolism: atypical findings from five cases affected by small molecule disorders

	J. Andres Morales, M.D. Pediatrics, Division of Medical Genetics, Stanford University, Stanford, CA
5:15 PM - 5:30 PM	The long non-coding RNA (LNCRNA) transcript *HULC* is regulator of the phenylalanine hydroxylase and a new potential treatment option for PKU patients Nenad Blau, Prof. Dr. (Ph.D.) Division of Metabolism, University Children's Hospital Zurich, Zurich, Switzerland
5:30 PM - 7:00 PM	Optional dinner for purchase (ticket required) otherwise dinner on own Butler – Level 1
Scientific Session 5 (7:00 PM- 10:00 PM)	Poster Session and Reception with Exhibits open Beer, Wine and Dessert served Panzacola F – Level 1
7:00 PM - 8:00 PM	Poster presentations from Travel Award Winners 5 min presentation at each poster from travel award winners (6 awards) attended by SIMD Founders Award Committee
7:00 PM - 7:10 PM Poster #1	Molecular and behavioral characterization of a novel mouse model of Snyder-Robinson syndrome: a path towards therapeutic development Oluwaseun Akinyele, Ph.D. Cancer Medicine, University of Pittsburgh, Pittsburgh, PA
7:10 PM - 7:20 PM Poster #2	Characterization of variants of uncertain significance in very long-chain Acyl-CoA dehydrogenase identified through newborn screening Olivia D'Annibale, Ph.D., M.P.H. University of Pittsburgh, Pittsburgh, PA
7:20 PM - 7:30 PM Poster #3	Predictors of survival in primary lactic acidosis Ibrahim Elsharkawi, M.D. Mitochondrial Medicine, Children's Hospital of Philadelphia, Philadelphia, PA
7:30 PM - 7:40 PM Poster #4	The current state of adult metabolic medicine in the United States: results of the first nationwide survey Jessica Gold, M.D., Ph.D. Children's Hospital of Philadelphia, Philadelphia, PA
7:40 PM - 7:50 PM Poster #5	Malate dehydrogenase deficiency: expanding the phenotypic spectrum Jessica R. C. Priestley, M.D., Ph.D. Children's Hospital of Philadelphia, Philadelphia, PA
7:50 PM - 8:00 PM Poster #6	Delayed skeletal development in a mouse model of global *SLC7A7* deficiency Bridget M. Stroup, Ph.D., R.D.N. Baylor College of Medicine, Houston, TX
8:00 PM - 8:30 PM	Committee review
7:00 PM - 9:00 PM	Posters attended by Authors 7:00-8:00 Even numbered posters attended 8:00-9:00 Odd numbered posters attended
Tuesday, April 12, 2022	
7:00 AM - 8:00 AM	Breakfast – Gatline Terrace – Level 1 Young Rare Disease Investigators Breakfast. Come and share YOUR experiences in rare disease research
7:30 AM - 1:00 PM	Meeting registration - Panzacola Registration – Level 1
Scientific Session 6 (8:00 AM -10:00 AM)	Title – IEMs as secondary findings: when does it matter? Panzacola G – Level 1 Moderators: Wendy Smith, M.D. and Kara Simpson, M.S., C.G.C. With the expansion of IEMs on the ACMG secondary finding Version 3 list, how should we change our approach to diagnosis and when to start management to avoid injury and avoid over-medicalization

8:00 AM - 8:20 AM	Secondary findings: rationale and approach to evaluation of genomically ascertained disorders of intermediary metabolism Les Biesecker, M.D. NIH Distinguished Investigator, Director, Center for Precision Health Research, Bethesda, MD
8:20 AM - 8:40 AM	How are gene-disease pairs added to the secondary findings list? David Miller, M.D. Assistant Professor of Pediatrics, Harvard Medical School, Boston, MA
8:40 AM - 9:05 AM	Fabry disease: Newborn screening and treatment implication Tamanna Roshan Lal, M.B.Ch.B. Children's National Hospital, Washington, DC
9:05 AM - 9:30 AM	The OTC example: What functional outcomes should lead to dietary changes Erin MacLeod, Ph.D. Director, Metabolic Nutrition, Children's National Hospital, Washington, DC
9:30 AM -10:00 AM	How can we use the secondary findings to expand adult onset metabolic disorders Moderators: Mark McNutt M.D., Ph.D., Tamanna Roshan Lal, M.B.Ch.B., Dawn Laney, M.S., C.G.C., C.C.R.C.
10:00 AM -11:00 AM	AM Coffee Break - Posters and Exhibits open - Panzacola F – Level 1
Scientific Session 7 11:00 AM -12:30 PM	Title – Biomarkers: Not everything worthwhile can be measured (and not everything measured is worthwhile) Panzacola G – Level 1 Moderators: Tatiana Yuzyuk, Ph.D. and Gerry Lipshutz, M.D.
11:00 AM -11:30 AM	Screening: GDF15 as a screening marker for mitochondrial disease Devin Oglesbee, Ph.D. Associate Professor, Laboratory Medicine and Pathology, Medical Genetics, Mayo Clinic, Rochester, MN
11:30 AM -12:00 PM	Disease Progression: Noninvasive biomarkers of liver disease in urea cycle disorders Lindsay Burrage, M.D., Ph.D. Assistant Professor, Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX
12:00 PM -12:30 PM	Diagnosis: Development of psychosine as a biomarker for Krabbe disease Dieter Matern, M.D., Ph.D. Mayo Clinic College of Medicine, Rochester, MN
12:30 PM -7:00 PM	Free afternoon - Lunch and dinner on your own
5:00 PM -7:00 PM	NAMA reception – Invitation ONLY – Lake Toho and Deck – Level 1
7:00 PM - 8:00 PM	Unknowns and Challenging Metabolic Cases Panzacola G – Level 1 Moderators: Jessie Priestley, M.D., Ph.D., Kirkland Wilson, M.D., Ph.D. and Debra Regier, M.D., Ph.D.
7:00 PM - 7:05 PM	Updates on old cases
7:05 PM - 8:00 PM	4-6 pre-submitted cases, 4 slides each presented by a moderator who has reviewed the cases ahead of time
8:00 PM - 9:30 PM	SIMD Award Presentations and Business Meeting Emmanuel Shapira SIMD Award (First Author of Best Publication in MGM) SIMD Founders Award (Best Oral Presentation by a Trainee)
Wednesday, April 13, 2022	
7:00 AM - 8:00 AM	Breakfast - Gatline Terrace – Level 1 SIMD interest groups: Nursing, Metabolic Nutrition, Genetic Counseling, Laboratorian, Basic Research, Clinical Research, Clinician

7:30 AM - 12:30 PM	Meeting Registration - Panzacola Registration – Level 1
Scientific Session 8 (8:00 AM - 10:00 AM)	Title: Fostering Cultural Humility in Current and Evolving Metabolic Practice Panzacola G – Level 1 Moderators: Erin MacLeod, Ph.D., R.D. and Suzanne Hollander, M.S., R.D.
8:00 AM - 8:30 AM	NBS and Culturally Competent Dietary Management from around the US and the World Barbara Marriage, Ph.D., R.D. Discovery, Product Research and Development, Abbott Nutrition, Columbus, OH and Rani Singh, Ph.D., R.D Professor in the Department of Human Genetics, Emory School of Medicine, Atlanta, GA
8:30 AM - 9:00 AM	Pseudodeficiencies in NBS for LSD Kerri Bosfield, MBBS Assistant Professor, Le Bonheur Children's Hospital, Memphis, TN
9:00 AM - 9:45 AM	Case presentations Clara Hildebrandt, M.D., University of North Carolina: Comprehensive Care for Ornithine Transcarbamylase Deficiency in A Family with Multiple Barriers to Care Laura Wagner, M.S., R.D., Children's Hospital of Michigan: MMA treatment for a family from Yemen Beth Ogata, M.S., R.D., CSP, University of Washington: Lessons learned working with a PKU family from Vietnam
10:00 AM -10:30 AM	AM coffee break – Posters and Exhibits open - Panzacola F – Level 1
Scientific Session 9 (10:30 AM - 12:30 PM)	Title: Cutting Edge Treatment Modalities Panzacola G – Level 1 Discussants: Cary Harding, M.D. and Irini Manoli, M.D., Ph.D.
10:30 AM - 11:15 AM	History of gene editing, technical and medicolegal aspects of CRISRP/Cas9 Wayne Grody, M.D., Ph.D. Professor in the Departments of Pathology & Laboratory Medicine, Pediatrics, and Human Genetics , UCLA, Los Angeles, CA
11:15 AM - 11:40 AM	Counseling families about genomic therapies for organic acidemias Jennifer Sloan, Ph.D. Staff Scientist, Molecular Geneticist and Genetic counselor NHGRI, NIH, Bethesda, MD
11:40 AM - 12:05 PM	In utero CRISPR therapeutic editing of metabolic genes William Peranteau, M.D. Assistant Professor of Surgery, Perelman School of Medicine, Children's Hospital of Pennsylvania, Philadelphia, PA
12:05 PM - 12:30 PM	mRNA for enzyme replacement in IEMs Gerald S Lipshutz, M.D., M.S. Professor in Residence, UCLA, Los Angeles, CA
12:30 PM - 12:45 PM	Looking Forward to SIMD 2023 and interim Events