

	SIMD 2019 April 6-9, 2019 Hyatt Regency, Bellevue Bellevue, Washington
Friday, April 5, 2019	
4:00 PM – 7:00 PM	Meeting Registration – Balsam Foyer
Saturday, April 6, 2019	
7:30 AM - 6:00 PM	Meeting Registration – Grand Ballroom Foyer
1:00 PM – 6:00 PM	Poster and Vendor set up – Evergreen Ballroom
7:00 AM – 8:30 AM	Breakfast on your own
9:15 AM	Shuttle bus leaves Hyatt Regency Hotel, Bellevue for ACMG meeting, Washington State Convention Center, Seattle
Scientific Session 1 (10:30 AM -12:00 AM)	Joint Session with ACMG Washington State Convention Center, 705 Pike Street, Seattle, WA You took the call from the Department of Health. What now? Moderators: Sue Berry, M.D. and Fernando Scaglia, M.D.
10:30 AM - 11:00 AM	<i>You Got the NBS Call for Presumptive Positive Pompe. What Now?</i> Priya Sunil Kishnani, M.D., M.B.B.S. Duke Department of Pediatrics Durham, NC
11:00 AM - 11:30 AM	<i>You Got the NBS Call for Presumptive Ppositive XALD. What Now?</i> Gerald V. Raymond, M.D. Penn State Hershey Medical Center Hershey, PA
11:30 AM - 12:00 AM	<i>You Got the NBS Call for Presumptive Positive MPS1. What Now?</i> Melissa P. Wasserstein, M.D. The Children’s Hospital at Montefiore Bronx, NY
12:00 PM -1:00 PM	Lunch - on your own – Washington State Convention Center
Scientific Session 2 (1:00 PM - 3:00 PM)	Joint Session with ACMG Washington State Convention Center, 705 Pike Street, Seattle, WA Could this really be metabolic? Dysmorphic features of IEMs Moderators: Gerry Berry, M.D. and Amarilis Sanchez-Valle, M.D.
1:00 PM - 1:30 PM	<i>Dysmorphism in Sterol Disorders</i> Forbes D. Porter, MD, PhD NICHD, NIH Rockville, MD
1:30 PM - 2:00 PM	<i>Dysmorphism in CDGs</i> Eva Morava-Kozicz, MD, PhD Mayo Clinic School of Medicine Rochester, MN
2:00 PM - 2:30 PM	<i>Dysmorphism in Mitochondrial Disorders</i> Rebecca Ganetzky MD

	Children's Hospital of Philadelphia Philadelphia, PA
2:30 PM - 3:00 PM	<i>Dysmorphism in Peroxisomal Disorders</i> Nancy Braverman, MD, MSc Research Institute-McGill University Health Center Montreal, Quebec
3:20 PM	Shuttle bus leaves Washington State Convention Center to return to the Hyatt Regency Hotel, Bellevue
3:15 PM – 6:00 PM	SIMD Board meeting – Alki Boardroom, 4 th floor Pike Street Tower Sheraton Seattle (ACMG Headquarters Hotel) 1400 6 th Ave, Seattle, WA
4:00 PM – 6:00 PM	NAMA BOD meeting – Cedar B (Hyatt Regency Bellevue)
7:00 PM – 10:00 PM	SIMD Opening Reception – Hyatt Regency Bellevue, Grand Ballroom A-D
Sunday, April 7, 2019	
7:00 AM – 8:00 AM	Breakfast – Grand Ballroom A-D 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 – Grand Ballroom Foyer
Scientific Session 3 (8:00 AM – 10:00 AM)	Plenary Talk – Grand Ballroom E-K Moderators: Gerry Berry, M.D. and Fernando Scaglia, M.D.
8:00 AM- 9:00 AM	<i>Elimination of Mutant Mitochondrial Genomes by Mitochondrial-Targeted DNA Editing Enzymes</i> Carlos T. Moraes, Ph.D. University of Miami, Miller School of Medicine Miami, FL
(9:00 AM- 12:00 PM)	Novel IEMs and treatments Moderators: Charles Venditti, M.D., Ph.D. and Penelope Bonnen, Ph.D.
9:00 AM – 9:30 AM	<i>PKAN Pathogenesis and Treatment</i> Susan Hayflick, M.D. Oregon Health & Science University Portland, OR
9:30 AM-10:00 AM	<i>MBTPS1 and SLC6A9 related disorders</i> Klaas Wierenga, M.D. Mayo Clinic Florida Jacksonville, FL
10:00 AM- 10:45 AM	AM coffee break – Posters and exhibits open– Evergreen Ballroom
Scientific Session 4	Challenges in Organ Transplantation for IEMs – Grand Ballroom E-K Moderator: Lynn Bush, Ph.D., M.S., M.A.
10:45 AM- 11:00 AM	<i>Clinical Case Vignette – The Drama of MMA: Medical Complexities and Challenges</i> Olaf Bodamer, M.D., Ph.D., Boston Children's Hospital, Boston, MA Lynn Bush, Ph.D, M.S.,M.A., Boston Children's Hospital and Columbia University Medical Center, Boston, MA Andrea Gropman, M.D., George Washington University, Children's National Medical Center, Washington, DC Laurie Smith, Ph.D., M.D., University of North Carolina School of Medicine, Chapel Hill, NC

	Charles Venditti, M.D., Ph.D., NHGRI/NIH, Bethesda, MD
11:00 AM-12:00 PM	<i>Panel Discussion – Treatment Decisions when the Data Are Not Clear and the Treatments Are Not Trivial</i> Carlos O. Esquivel, M.D., Ph.D. The Arnold and Barbara Silverman Professor of Surgery, Chief of the Division Abdominal Transplantation Stanford School of Medicine Stanford, CA
12:00 PM -1:00 PM	Lunch - boxed lunch – Grand Ballroom A-D
Scientific Session 5 (1:00 PM - 3:00 PM)	Oral presentations from Travel Award winners, Shapira Award winner and the JSIMD Young Investigator winner – Grand Ballroom E-K Moderators: Shawn E. McCandless, M.D. and Sarah Viall, B.S., M.S.N.
1:00 PM - 1:15 PM	<i>Multi-Omics Studies in Patient-Derived and CRISPR-Edited Cellular Models of Methylmalonic Acidemia and Propionic Acidemia Reveal Dysregulation of Serine Metabolism: New Directions for Cellular Pathogenesis in Disorders of Branch Chain Amino Acid Metabolism</i> Arianna Franca Anzmann, M.S. McKusick-Nathans Institute of Genetic Medicine, Johns Hopkins School of Medicine Baltimore, MD
1:15 PM - 1:30 PM	<i>A Network View of VLCAD Deficiency: From Common to Rare Disease Biology</i> Aaron Bender, B.A. Icahn School of Medicine at Mount Sinai New York, NY
1:30 PM - 1:45 PM	<i>Biomarkers of Chronic Kidney Disease in Methylmalonic Acidemia (MMA): Monitoring and Treatment Ramifications</i> Carolina Isabel Galarreta Aima. M.D. NHGRI/NIH Bethesda, MD
1:45 PM - 2:00 PM	<i>Successful CRISPR/Cas9-Mediated Gene Editing in Murine Phenylketonuria (PKU)</i> Daelyn Richards, Ph.D. Candidate Oregon Health & Science University Portland, OR
2:00 PM - 2:15 PM	<i>Early Diagnosis, Diet and Cofactor Therapy Affect Neurological Outcomes in Cobalamin C (cblC) Deficiency</i> Sho Yano, M.D., Ph.D. National Human Genome Research Institute, NIH Bethesda, MD
2:15 PM - 2 :30 PM	<i>Treatment of Bioenergetic Dysfunction In Propionic Acidemia</i> Zetong Zheng University of Pittsburgh, School of Medicine Pittsburgh, PA
2:30 PM – 2:45 PM	Presentations from the 2018 Emmanuel Shapira Award Winner TBD
2:45 PM – 3:00 PM	JSIMD Young Investigator award winner for best presentation at 2018 JSIMD meeting

	<i>Understanding Pathomechanisms of Lysosomal Storage Disorders with Cellular Models Using Genome-Editing Technology</i> Takanobu Otomo, M.D., Ph.D. Kawasaki Medical School Okayama, Japan
3:00 PM - 4:00 PM	PM coffee break – Posters and exhibits open – Evergreen Ballroom
Scientific Session 6 (4:00 PM- 5:30 PM)	Platform Presentations - Grand Ballroom E-K Selected Abstracts presentations Moderators: Susan Berry, M.D. and Amy White, M.S., C.G.C.
4:00 PM - 4:15 PM	<i>Sustained Reductions in Plasma Arginine Following Pegzilarginase Administration in Patients with Arginase-1 Deficiency are Accompanied by Improvements in Mobility and Adaptive Behavior</i> George Diaz, M.D., Ph.D. Icahn School of Medicine at Mount Sinai New York, NY
4:15 PM - 4:30 PM	<i>Neurological Phenotype in Methylmalonic Acidemia Transgenic Mice in the Well and Decompensated State</i> Jamie Fraser, M.D., Ph.D. Rare Disease Institute, Children’s National Medical Center Washington, DC
4:30 PM - 4:45 PM	<i>A Phase 1/2 Clinical Trial for AAV8-Mediated Liver-Directed Gene Therapy in Adults with Late-Onset OTC Deficiency: Interim Results from Cohort 1</i> Cary O. Harding, M.D. Oregon Health & Science University Portland, OR
4:45 PM - 5:00 PM	<i>High Levels of Mitochondrial DNA Heteroplasmy with 3243A>G Mutation in Human iPSC-Derived Excitatory Cortical Neurons Causes Reduced Neuronal Activity and Altered Neuronal Network Organization</i> Tamás Kozicz M.D., Ph.D. Mayo Clinic Rochester, MN
5:00 PM - 5:15 PM	TBD
5:15 PM - 5 :30 PM	TBD
5:30 PM – 7:00 PM	Dinner – ticket required – Grand Ballroom A-D
Scientific Session 7 (7:15 PM – 10:00 PM)	Poster Session and Reception with Exhibits open- Evergreen Ballroom
7:30 PM – 8:45 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
7:30 PM - 7:40 PM Poster #1	<i>Metabolomic Comparison of Triheptanoin and Medium Chain Triglyceride Therapy in Very-Long-Chain Acyl-CoA Dehydrogenase Deficiency</i> Ahmad Alodaib, Ph.D. Children’s Hospital of Pittsburgh, Pittsburgh, PA
7:40 PM - 7:50 PM Poster #2	<i>The Metabolic Map of the Pathomechanism and Treatment of PGM1-CDG</i> Matthew Bird, Ph.D. Metabolomics Expertise Center, VIB & Hepatology Laboratory - KUL Leuven, Belgium

7:50 PM - 8:00 PM Poster #3	<i>IVD Deficient Fibroblasts Show Improvement in Phenotype with Epigallocatechin Gallate Treatment</i> Olivia D'Annibale, MPH University of Pittsburgh Pittsburgh, PA
8:00 PM - 8:10 PM Poster #4	<i>Changing the Clinical Course of Infantile Pompe Disease with Immune Modulation Strategies: 12 Years of Experience</i> Ankit Desai, M.B.B.S. Duke University Medical Center Durham, NC
8:10 PM - 8:20 PM Poster #5	<i>Liver Transplantation in Mitochondrial Neurogastrointestinal Encephalomyopathy (MNGIE)</i> Kimberly Kripps, M.D. University of Colorado Denver and Children's Hospital of Colorado Denver, CO
8:20 PM - 8:30 PM Poster #6	<i>Pharmacologic Rescue of Hyperammonemia-Induced Neurotoxicity by Inhibition of Ornithine Aminotransferase in a Zebrafish Model of Acute Hyperammonemic Encephalopathy</i> Matthias Zielonka, M.D. University Hospital Heidelberg, Center for Child and Adolescent Medicine Heidelberg, Germany
8:30 PM - 8:45 PM	Committee review
Monday, April 8, 2019	
7:00 AM - 8:00 AM	Breakfast – Grand Ballroom A-D Young Rare Disease Investigators Breakfast. Come and share YOUR experiences in rare disease research
8:00 AM - 1:00 PM	Meeting registration – Grand Ballroom Foyer
Scientific Session 8 (8:00 AM - 10:00 AM)	Diagnosis of IEMs: Complementary Use of Genomics and Metabolomics - Grand Ballroom E-K Moderators: Gerry Berry, M.D. and Marzia Pasquali, Ph.D.
8:00 AM - 8:20 AM	<i>The Confluence of Genomic Sequencing and Newborn Screening</i> Cindy Powell, M.D. The University of North Carolina at Chapel Hill Chapel Hill, NC
8:20 AM - 8:40 AM	<i>Standards-Free Metabolomics: Shifting the Paradigm Through Advanced Computation and Instrumentation</i> Tom O. Metz, Ph.D. Pacific Northwest National Laboratory Richland, WA
8:40 AM - 9:00 AM	<i>Pros and Cons of Metabolomics vs Targeted Testing</i> Devin Oglesbee, Ph.D. Mayo Clinic Rochester, MD
9:00 AM - 9:20 AM	<i>Augmenting the Use of Metabolomics and Genomics Testing</i> Sarah H. Elsea, Ph.D. Baylor College of Medicine Houston, TX

9:20 AM - 9:40 AM	<i>Metabolomics as a Diagnostic Tool: Lessons from the UDN</i> David M. Koeller, M.D. Oregon Health & Science University Portland, OR
9:40 AM - 10:00 AM	Discussion
10:00 AM -11:00 AM	AM Coffee Break - Posters and Exhibits open – Evergreen Ballroom
Scientific Session 9 (11:00 AM - 12:30 PM)	Improving Patient Care in the 21st Century Moderators: Georgianne Arnold, M.D. and Wendy Smith, M.D.
11:00 AM -11:30 AM	<i>Creation of Evidence and Consensus-Based Nutrition Guidelines in IMD with Limited Research</i> Fran Rohr, M.D., Boston Children’s Hospital, Boston, MA and Suzanne Hollander, M.S., R.D., UCLA Health, Los Angeles, CA
11:30 AM -12:30 PM	<i>Quality Improvement Research: Advancing Metabolic Care for all of our Patients</i> Amy Kritzer, M.D., Boston Children’s Hospital, Boston, MA and Debra Regier, M.D., Ph.D., Children’s National Medical Center, Washington, DC
12:30 PM -7:00 PM	Free afternoon - Lunch and dinner on your own
1:30 PM – 5:30 PM	Cerebral Creatine Deficiency Syndrome (CCDS) Workshop Sponsored by the Association for Creatine Deficiencies Grand Ballroom E-K
5:00 PM -7:00 PM	NAMA reception – Invitation ONLY – Grand Ballroom A-D
7:00 PM – 7:15 PM	Unknown case – Grand Ballroom E-K Updates on old cases - Georgianne Arnold, M.D. and Debra Regier, M.D., Ph.D.
7:15 PM - 7:45 PM	2-3 pre-submitted cases, 4 slides each presented by a moderator who will be asked to review ahead of time
7:45 PM - 8:15 PM	Presidential Address Update from the Workgroups of the SIMD Clinical Research Network Shawn E. McCandless, M.D., SIMD President University of Colorado Denver Aurora, CO
8:15 PM - 9:30 PM	SIMD Business Meeting and Award Presentations Emmanuel Shapira SIMD Award (First Author of Best Publication in MGM) SIMD Founders Award (Best Oral Presentation by a Trainee)
Tuesday, April 9, 2019	
7:00 AM -8:00 AM	Breakfast – Grand Ballroom A-D Clinical Experts Breakfast. Join a table of YOUR favorite clinical disorder and chat about YOUR patients or YOUR questions
7:30 AM - 12:00 PM	Meeting Registration – Grand Ballroom Foyer
Scientific Session 10 (8:00 AM - 10:00 AM)	Treatment challenges for VLCAD deficiency – Grand Ballroom E-K Moderators: Jerry Vockley, M.D. and Rani Singh, Ph.D., R.D., L.D.
8:00 AM - 8:30 AM	<i>GMDI/SERN Nutrition Management Guidelines for VLCAD: What’s the Evidence?</i> Sandy Van Calcar, Ph.D., R.D., L.D. Oregon Health & Science University Portland, OR

8:30 AM - 9:00 AM	<i>Is Low Carnitine a Problem? To Supplement or Not to Supplement.</i> Curtis Coughlin, M.S., MBe University of Colorado, Children's Hospital of Colorado Aurora, CO
9:00 AM - 9:30 AM	<i>What Do Plasma Acylcarnitines Tell Us?</i> Melanie Gillingham, Ph.D. Oregon Health & Science University Portland, OR
9:30 AM -10:00 AM	<i>Positive NBS for VLCAD; Now What? The Dutch Approach</i> Gepke Visser, M.D., Ph.D. UMCU Utrecht, The Netherlands
10:00 AM -10:30 AM	AM coffee break – Posters and Exhibits open - Evergreen Ballroom
Scientific Session 11 (10:30 AM - 12:00 PM)	Eye in IEMs – Grand Ballroom E-K Moderators: Laurie Smith, Ph.D., M.D., and Irini Manoli, M.D., Ph.D.
10:30 AM - 11:00 AM	<i>LCHADD Retinopathy</i> Mark Pennesi, M.D., Ph.D. Casey Eye Institute, Oregon Health & Science University Portland, OR
11:00 AM - 11:30 AM	<i>Ophthalmic Abnormalities in Patients with CblC and Isolated MMA</i> Brian P. Brooks, M.D., Ph.D. National Eye Institute Bethesda, MD
11:30 AM - 12:00 PM	TBD
12:00 PM – 12:15 PM	Closing comments – END OF MEETING