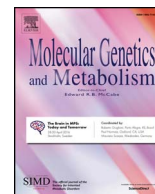


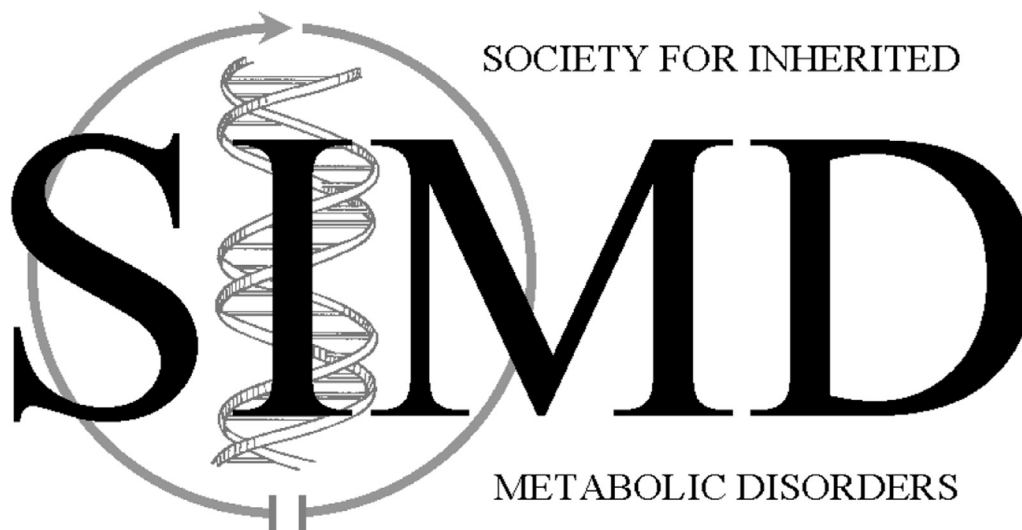


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Abstracts



Society for Inherited Metabolic Disorders
40th Annual Meeting

Sunday, March 11, 2018 – Wednesday, March 14, 2018
Paradise Point
San Diego, CA

Program and Abstracts

Program for the 2018 SIMD Annual Meeting

Sunday, March 11, 2018

- 1:00 PM – 6:00 PM Meeting Registration – Paradise Foyer
 1:00 PM – 6:00 PM Poster board and vendor set up – Sunset Pavilion
 3:00 PM – 6:00 PM SIMD Board meeting – Executive Suites, Rm# 713
 7:00 PM – 10:00 PM Opening Reception – **Paradise Terrace**

Monday, March 12, 2018

- 7:30 AM - 5:00 PM Meeting registration - **Paradise Foyer**
 7:00 AM - 8:00 AM **Breakfast – Paradise Terrace**
 New to SIMD, new to Rare Disease, or want to meet for breakfast? Join us at the tables labeled “New to SIMD”

Scientific Session 1 Politics, Economics & the Treatment of IEM's

(8:00 AM -10:00 AM) Paradise Ballroom

Moderators: Laurie D. Smith, Ph.D., M.D. & Debra Regier, M.D., Ph.D.

8:00 AM - 8:15 AM Introduction and Welcome

Gerard Berry, M.D., 2018 Program Chairman

8:15 AM - 8:45 AM Cost as a Barrier to Accessing New Treatments for Rare Diseases: Economic Perspectives

Scott D. Grosse, Ph.D.

National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, GA

8:45 AM - 9:30 AM Discussion on Treatment of Rare IMDs and the Role of the SIMD as an Advocate

9:30 AM -10:00 AM 7th Robert Guthrie Memorial Lecture

What are the Limits of Newborn Screening?

Susan A. Berry, M.D.

University of Minnesota, Minneapolis, MN

10:00 AM-11:00 AM AM Coffee Break-Posters and Exhibits open - **Sunset Pavilion**

Scientific Session 2 Congenital Disorders of GlycosylationParadise Ballroom

(11:00 PM - 12:00 PM) Moderator: Gerard Berry, M.D.

11:00 AM - 12:00PM Congenital Disorders of Glycosylation: Impaired Fucosylation and Changing Perspectives

Hudson Freeze, Ph.D.

Sanford-Burnham-Prebys Medical Discovery Institute, La Jolla, CA

12:00 PM -1:15 PM Box Lunch - **Paradise Terrace**

Scientific Session 3 Travel Award and Shapira Award Presentations

(1:15 PM – 3:00 PM) Paradise Ballroom

Moderators: Cate Walsh Vockley, M.S., L.C.G.C. & Irini Manoli, M.D., Ph.D.

1:15 PM - 1:30 PM Bezaifibrate Treatment Enhances Efficiency of AAV Gene Therapy on Glycogen Storage Disease Type IA

Hye Ri Kang, Ph.D.

Duke University, Durham, NC

1:30 PM - 1:45 PM New Mitochondrial Diagnostic Testing in Fibroblasts, A Minimally Invasive Tissue,

Identifies Sensitive Functional Testing Methods For Complex I Deficient Primary Mitochondrial Disorders

Kaz Knight, B.S.

University of Colorado, Anschutz Medical Campus, Aurora, CO

1:45 PM - 2:00 PM Nucleoside Bioavailability and Tk1 Steady-State Levels Modulate Response to dC + dT

Therapy in Mitochondrial DNA Depletion Syndrome Caused by TK2 Deficiency

Carlos Lopez-Gomez, Ph.D.

H. Houston Merritt Center for Neuromuscular Disorders, Columbia University Medical Center, New York, NY

2:00 PM - 2:15 PM Mitochondrial Targeted Antioxidants Improve Mitochondrial Function in Very Long-Chain Acyl-CoA Dehydrogenase Deficient Fibroblasts

Bianca Seminotti, Ph.D.

University of Pittsburgh, Pittsburgh, PA & PPG Ciências Biológicas: Bioquímica, Universidade Federal do Rio Grande do Sul, Brazil

2:15 PM - 2:30 PM Interference of Tricarboxylic Acid Cycle Enzyme Activities in Methylmalonyl-CoA

Mutase Knock-out/Knock-in Mice

Parith Wongkittichote, Ph.D., M.D.

Children's National Medical Center, Washington, DC

2:30 PM - 2:45 PM Development of a Minimally Invasive Muscle Oxygen Nanosensor for Evaluation of Mitochondrial Myopathy

Zarazuela Zolkipli-Cunningham, M.B., Ch.B., M.R.C.P.

The Children's Hospital of Philadelphia, Philadelphia, PA

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