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|                                                                                   | <hr/> <h1>SIMD 2018</h1> <p><b>March 11-14, 2018</b><br/> <b>Paradise Point</b><br/> <b>San Diego, California</b></p>                                                                                                                     |
| <b>Sunday, March 11, 2018</b>                                                     |                                                                                                                                                                                                                                           |
| 1:00 PM – 6:00 PM<br>1:00 PM – 6:00 PM<br>3:00 PM – 6:00 PM<br>7:00 PM – 10:00 PM | Meeting Registration – Paradise Foyer<br>Poster board and vendor set up – Sunset Pavilion<br>SIMD Board meeting – Executive Suites, Rm# 713<br>Opening Reception – <b>Paradise Terrace</b>                                                |
| <b>Monday, March 12, 2018</b>                                                     |                                                                                                                                                                                                                                           |
| 7:30 AM - 5:00 PM                                                                 | Meeting registration - <b>Paradise Foyer</b>                                                                                                                                                                                              |
| 7:00 AM - 8:00 AM                                                                 | <b>Breakfast – Paradise Terrace</b><br>New to SIMD, new to Rare Disease, or want to meet for breakfast? Join us at the tables labeled “New to SIMD”                                                                                       |
| <b>Scientific Session 1</b><br>(8:00 AM -10:00 AM)                                | <b>Politics, economics &amp; the treatment of IEMs</b><br><b>Paradise Ballroom</b><br>Moderators: Laurie D. Smith, Ph.D., M.D. & Debra Regier, M.D., Ph.D.                                                                                |
| 8:00 AM - 8:15 AM                                                                 | <i>Introduction and Welcome</i><br>Gerard Berry, M.D., 2018 Program Chairman                                                                                                                                                              |
| 8:15 AM - 8:45 AM                                                                 | <i>Economic Perspectives on Rare Diseases, Orphan Drugs, And Diagnostic Utility</i><br>Scott D. Grosse, Ph.D.<br>National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention, Atlanta, GA |
| 8:45 AM - 9:30 AM                                                                 | <i>Point-Counterpoint Discussion Between Health Policy Advisors</i><br>Karen L. Summar, M.D., M.S.<br>Health Policy Director, U.S. Senator Chuck Grassley<br>Washington, DC                                                               |
| 9:30 AM -10:00 AM                                                                 | <b>7<sup>th</sup> Robert Guthrie Memorial Lecture</b><br><i>What are the Limits of Newborn Screening?</i><br>Susan A. Berry, M.D.<br>University of Minnesota, Minneapolis, MN                                                             |
| 10:00 AM-11:00 AM                                                                 | AM Coffee Break-Posters and Exhibits open - <b>Sunset Pavilion</b>                                                                                                                                                                        |
| <b>Scientific Session 2</b><br>(11:00 PM - 12:00 PM)                              | <b>Congenital Disorders of Glycosylation</b><br><b>Paradise Ballroom</b><br>Moderator: Gerard Berry, M.D.                                                                                                                                 |
| 11:00 AM - 12:00PM                                                                | <i>Congenital Disorders of Glycosylation: Impaired Fucosylation and Changing Perspectives</i><br>Hudson Freeze, Ph.D.<br>Sanford-Burnham-Prebys Medical Discovery Institute, La Jolla, CA                                                 |
| 12:00 PM -1:15 PM                                                                 | Box Lunch - <b>Paradise Terrace</b>                                                                                                                                                                                                       |
| <b>Scientific Session 3</b><br>(1:15 PM – 3:00 PM)                                | <b>Travel Award Presentations</b><br><b>Paradise Ballroom</b><br>Moderators: Cate Walsh Vockley, M.S., L.C.G.C. & Irini Manoli, M.D., Ph.D.                                                                                               |
| 1:15 PM - 1:30 PM                                                                 | <i>Bezafibrate Treatment Enhances Efficiency of AAV Gene Therapy on Glycogen Storage Disease Type IA</i><br>Hye Ri Kang, Ph.D.<br>Duke University, Durham, NC                                                                             |

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| 1:30 PM - 1:45 PM                                 | <i>New Mitochondrial Diagnostic Testing in Fibroblasts, A Minimally Invasive Tissue, Identifies Sensitive Functional Testing Methods For Complex I Deficient Primary Mitochondrial Disorders</i><br>Kaz Knight, B.S.<br>University of Colorado, Anschutz Medical Campus, Aurora, CO                       |
| 1:45 PM - 2:00 PM                                 | <i>Nucleoside Bioavailability and Tk1 Steady-State Levels Modulate Response to dC+dT Therapy in Mitochondrial DNA Depletion Syndrome Caused by TK2 Deficiency</i><br>Carlos Lopez-Gomez, Ph.D.<br>H. Houston Merritt Center for Neuromuscular Disorders, Columbia University Medical Center, New York, NY |
| 2:00 PM - 2:15 PM                                 | <i>Mitochondrial Targeted Antioxidants Improve Mitochondrial Function in Very Long-Chain acyl-CoA Dehydrogenase Deficient Fibroblasts</i><br>Bianca Seminotti, Ph.D.<br>University of Pittsburgh, Pittsburgh, PA & PPG Ciências Biológicas: Bioquímica, Universidade Federal do Rio Grande do Sul, Brazil |
| 2:15 PM - 2:30 PM                                 | <i>Interference of Tricarboxylic Acid Cycle Enzyme Activities in Methylmalonyl-CoA Mutase Knock-out/Knock-in Mice</i><br>Parith Wongkittichote, Ph.D., M.D.<br>Children's National Medical Center, Washington, DC                                                                                         |
| 2:30 PM - 2:45 PM                                 | <i>Development of a Minimally Invasive Muscle Oxygen Nanosensor for Evaluation of Mitochondrial Myopathy</i><br>Zarazuela Zolkipl- Cunningham, M.B., Ch.B., M.R.C.P.<br>The Children's Hospital of Philadelphia, Philadelphia, PA                                                                         |
| 2:45 PM – 3:00 PM                                 | <i>Shapira award winner presentation –TBD</i>                                                                                                                                                                                                                                                             |
| 3:00 PM - 3:30 PM                                 | PM Coffee break – Posters and Exhibits open - <b>Sunset Pavilion</b>                                                                                                                                                                                                                                      |
| <b>Scientific Session 4</b><br>(3:30 PM – 5:20PM) | <b>Selected abstract presentations and JSIMD Young Investigators</b><br><b>Paradise Ballroom</b><br><b>Moderators: Marzia Pasquali, Ph.D. &amp; Penelope Bonnen, Ph.D.</b>                                                                                                                                |
| 3:30 PM – 3:45 PM                                 | <i>Prevalences and Mechanisms of Chronic Hepatic Complications in Urea Cycle Disorders</i><br>Lindsay C. Burrage, M.D., Ph.D. <b>(2018 Travel Award Winner)</b><br>Baylor College of Medicine, Houston, TX                                                                                                |
| 3:45 PM – 4:00 PM                                 | <i>Correction of Murine Models of Methylmalonic Acidemia using Albumin Targeted Homologous Recombination with a Promoterless Adeno-Associated Viral Integrating Vector</i><br>Randy J. Chandler, Ph.D., M.B.<br>National Human Genome Research Institute, National Institutes of Health Bethesda, MD      |
| 4:00 PM - 4:15 PM                                 | <i>Mutations in the Genes QRSL1, GATB, And GATC Encoding the Subunits of Glutamyl-tRNA<sup>gln</sup> Amidotransferase Cause a Mitochondrial Disorder with Lethal Infantile Cardiomyopathy</i><br>Marisa W. Friederich, Ph.D. <b>(2018 Travel Award Winner)</b><br>University of Colorado, Aurora, CO      |
| 4:15 PM - 4:30 PM                                 | <i>Immunotherapy Alleviates Metabolic Decompensation Due to Infection in a Mouse Model of Mitochondrial Hepatopathy</i><br>Maxim Jestin, B.S. <b>(2018 Travel Award Winner)</b><br>National Human Genome Research Institute, National Institutes of Health Bethesda, MD                                   |
| 4:30 PM - 4:45 PM                                 | <i>First Reported Pediatric Case of Glucagon Receptor Dysfunction Identified Through Newborn Screening</i>                                                                                                                                                                                                |

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|                                                    | Hong Li, M.D., Ph.D.<br>Emory University School of Medicine, Atlanta, GA                                                                                                                                                                                                                                        |
| 4:45 PM – 5:00 PM                                  | <i>Modeling Hepatic Stress Adaptation in Methylmalonic Acidemia</i><br>Irin Manoli, M.D., Ph.D.<br>National Human Genome Research Institute, National Institutes of Health Bethesda, MD                                                                                                                         |
| 5:00 PM – 5:10 PM                                  | JSIMD Young Investigator for best presentation at the 2016 JSIMD meeting<br><i>Identification of Biallelic Mutations in IARS, a Known Causative Gene of Weak Calf Syndrome, which Cause Infantile Mitochondrial Hepatopathy</i><br>Masaru Shimura, M.D.<br>Chiba Children's Hospital, Chiba, Japan              |
| 5:10 PM - 5:20PM                                   | JSIMD young investigator for best presentation at the 2017 JSIMD meeting<br><i>Mitochondrial Acid 5 (MA-5) Facilitates ATP Synthase Oligomerization and Cell Survival in Various Mitochondrial Diseases</i><br>Tetsuro Matsushashi, M.D., Ph.D.<br>Tohoku University Graduate School of Medicine, Sendai, Japan |
| 5:45 PM - 7:00 PM                                  | Dinner on own <b>OR</b> Optional dinner for purchase (ticket required) – Mission Bay Room                                                                                                                                                                                                                       |
| <b>Scientific Session 5</b><br>(7:00 PM- 10:00 PM) | <b>Presidential Talk and Poster Session</b>                                                                                                                                                                                                                                                                     |
| 7:00 PM - 7:30 PM                                  | <b>Presidential Address</b><br><b>Paradise Ballroom</b><br><i>Ways of Knowing, Evidenced-Based Medicine and the SIMD</i><br>Shawn McCandless, M.D., SIMD President<br>Case Western Reserve University, Cleveland, OH<br>Introduction: <i>Gerard Berry, M.D.</i>                                                 |
| 7:30 PM -10:00 PM                                  | <b>Posters attended by Authors and Exhibits open</b><br><b>Sunset Pavilion</b>                                                                                                                                                                                                                                  |
|                                                    | <b>Beer, Wine and Dessert served</b>                                                                                                                                                                                                                                                                            |
|                                                    | <b>7:30-8:30 Even numbered posters attended</b>                                                                                                                                                                                                                                                                 |
|                                                    | <b>8:30-9:30 Odd numbered posters attended</b>                                                                                                                                                                                                                                                                  |
| 7:30 PM – 7:55 PM                                  | <b>Travel Award Poster Presentations</b><br>5 min presentation at each poster from 3 travel award winners attended by SIMD Founders Award Committee                                                                                                                                                             |
| 7:30 PM – 7:35 PM<br>Poster #1                     | <i>Expression Pattern of Phenylalanine Hydroxylase Variants is Regulated by Co-Chaperone DNAJC12</i><br>Nastassja Himmelreich, Ph.D.<br>University Children's Hospital, Dietmar-Hopp-Metabolic Center, Heidelberg, Germany                                                                                      |
| 7:40 PM – 7:45 PM<br>Poster #2                     | <i>A Novel RMND1 Knock-In Mouse Recapitulates Clinical and Biochemical Abnormalities Observed in Patients</i><br>Maria Sanchez-Quintero, Ph.D.<br>Columbia University Medical Center, New York, NY                                                                                                              |
| 7:50 PM – 7:55 PM<br>Poster #3                     | <i>Characterization of a New Nonketotic Hyperglycinemia Mouse Model with An Unstable Missense Mutation In GLDC</i><br>Michael A. Swanson, Ph.D., University of Colorado, Denver, CO                                                                                                                             |
| <b>Tuesday, March 13, 2018</b>                     |                                                                                                                                                                                                                                                                                                                 |
| 7:00 AM - 8:00 AM                                  | <b>Breakfast – Paradise Terrace</b><br>Young Rare Disease Investigators Breakfast. Come and share YOUR experiences in rare disease research                                                                                                                                                                     |
| 7:30 AM - 1:00 PM                                  | Meeting registration - <b>Paradise Foyer</b>                                                                                                                                                                                                                                                                    |

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| <b>Scientific Session 6</b><br>(8:00 AM - 10:00 AM)  | <b>Genomic Therapies in IEM</b><br><b>Paradise Ballroom</b><br>Moderators: Charles Venditti, M.D., Ph.D. & Penelope Bonnen, Ph.D.                                                                                                               |
| 8:00 AM - 8:30 AM                                    | <i>Overview of Nucleic Acid Therapies</i><br>Dwight D. Koeberl, M.D., Ph.D.<br>Duke University Hospital, Durham, NC                                                                                                                             |
| 8:30 AM - 9:00 AM                                    | <i>Design of an Open Label Phase 1/2 Clinical Trial for AAV8-Mediated Liver-Directed Gene Therapy in Adults with Late Onset Ornithine Transcarbamylase Deficiency</i><br>Cary Harding, M.D.<br>Oregon Health & Science University, Portland, OR |
| 9:00 AM - 9:30 AM                                    | <i>Gene Therapies for the Mucopolysaccharidoses</i><br>Joseph Muenzer, M.D., Ph.D.<br>University of North Carolina at Chapel Hill, Chapel Hill, NC                                                                                              |
| 9:30 AM - 10:00 AM                                   | <i>Gene Therapy for the CNS Manifestations of Hereditary Disorders</i><br>Ronald G. Crystal, M.D.<br>Weill Cornell Medical College, New York, NY                                                                                                |
| 10:00 AM -11:00 AM                                   | <b>AM Coffee Break - Posters and Exhibits open – Sunset Pavilion</b>                                                                                                                                                                            |
| <b>Scientific Session 7</b><br>(11:00 AM - 12:30 PM) | <b>Updated dietary treatment guidelines</b><br><b>Paradise Ballroom</b><br>Moderators: Melanie Gillingham, Ph.D. & Kathy Camp, M.D., R.D.                                                                                                       |
| 11:00 AM -11:30 AM                                   | <i>Protein Intake Recommendations for Propionic Acidemia in the Evidence-based SERN-GMDI Management Guidelines</i><br>Elaina Jurecki, M.S., R.D.<br>BioMarin Pharmaceuticals, San Rafael, CA                                                    |
| 11:30 AM -12:00 PM                                   | <i>GA1 Guideline and Nutrition Management</i><br>Krista Viau, Ph.D., R.D.N., C.S.P.<br>Boston Children's Hospital, Boston, MA                                                                                                                   |
| 12:00 PM - 12:30 PM                                  | <i>Glycomacropeptide for Nutritional Management of Phenylketonuria</i><br>Denise M. Ney, Ph.D., R.D.N.<br>University of Wisconsin – Madison, Madison, WI                                                                                        |
| 1:00 PM – 2:00 PM                                    | <b>Elsevier Workshop</b> - Paradise Ballroom<br><i>Scholarly Publishing Pearls &amp; Pitfalls: Maximizing the Potential of Your Next Submission</i><br>David Parson, Publisher, Elsevier<br>New York, NY                                        |
| 12:30 PM -7:00 PM                                    | <b>Free afternoon - Lunch and dinner on your own</b>                                                                                                                                                                                            |
| 5:00 PM - 7:00 PM                                    | <b>NAMA reception – Invitation ONLY - Island Point Lawn</b>                                                                                                                                                                                     |
| <b>Scientific Session 8</b><br>(7:00 PM - 8:00 PM)   | <b>Unknowns and Challenging Metabolic Cases</b><br><b>Paradise Ballroom</b><br>Moderators: Amarilis Sanchez-Valle, M.D., Georgianne Arnold, M.D., Debra Regier, M.D., Ph.D.                                                                     |
| 7:00 PM – 7:05 PM                                    | Updates on old cases                                                                                                                                                                                                                            |
| 7:05 PM – 8:00 PM                                    | Six pre-submitted cases, 4 slides each presented by a moderator who will review ahead of time                                                                                                                                                   |
| 8:00 PM - 9:30 PM                                    | <b>SIMD Business Meeting and Award Presentations</b><br><b>Paradise Ballroom</b><br>Emmanuel Shapira SIMD Award (First Author of Best Publication in MGM)<br>SIMD Founders Award (Best Oral Presentation by a Trainee)                          |
| <b>Wednesday, March 14, 2018</b>                     |                                                                                                                                                                                                                                                 |

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| 7:00 AM - 8:00 AM                                     | <b>Breakfast – Paradise Terrace</b><br>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 - <b>Paradise Foyer</b>                                                                                                                                                          |
| <b>Scientific Session 9</b><br>(8:00 AM - 9:00 AM)    | <b>The Rare Disease Consortia</b><br><b>Paradise Ballroom</b><br>Moderators: Amarilis Sanchez-Valle, MD. & Cindy Freehauf, R.N., M.S., C.G.C.                                                         |
| 8:00 AM - 8:30 AM                                     | <i>Urea Cycle Disorders Consortium Highlights</i><br>Sandesh C.S. Nagamani, M.D., FACMG<br>Baylor College of Medicine, Houston, TX                                                                    |
| 8:30 AM - 9:00 AM                                     | <i>Peroxisomal Disease Global Foundation: Physicians, Scientists and Families Working Together</i><br>Nancy Braverman, M.D., M.Sc., FACMG<br>McGill University Health Centre, Montreal, Quebec Canada |
| <b>Scientific Session 10</b><br>(9:00 AM - 11:00 AM)  | <b>Mitochondrial Metabolism – Paradise Ballroom</b><br>Moderators: Fernando Scaglia, M.D., & Shawn McCandless, M.D.                                                                                   |
| 9:00 AM - 9:30 AM                                     | <i>One Gene, Many Faces: The Many Phenotypes Of POLG Disease</i><br>Robert Naviaux, M.D., Ph.D.<br>University of California, San Diego, San Diego, CA                                                 |
| 9:30 AM - 10:00 AM                                    | <i>Nucleoside Therapy for Disorders of Mitochondrial DNA Maintenance</i><br>Michio Hirano, M.D.<br>Columbia University, New York, NY                                                                  |
| 10:00 AM - 10:30 AM                                   | <i>Mitochondrial Fusion and Fission in Health and Disease</i><br>David Chan, Ph.D.<br>California Institute of Technology, Pasadena, CA                                                                |
| 10:30 AM - 11:00 AM                                   | <i>Repurposing Screening Identifies Drugs that Provide Benefit in Cell and Animal Models of Mitochondrial Disease</i><br>Gino Cortopassi, Ph.D.<br>University of California, Davis, Davis, CA         |
| 11:00 AM - 11:30 AM                                   | AM coffee break – Posters and Exhibits open – <b>Sunset Pavilion</b>                                                                                                                                  |
| <b>Scientific Session 11</b><br>(11:30 AM - 12:30 PM) | <b>Genomic Aspects of IEM</b><br>Moderators: Klaas Wierenga, M.D. & Laurie D. Smith. Ph.D., M.D.                                                                                                      |
| 11:30 AM - 12:00 PM                                   | <i>Genome Wide Sequencing in Neonates Facilitates Rapid Specific Diagnosis</i><br>David Dimmock, M.D.<br>Rady Children's Institute for Genomic Medicine, San Diego, CA                                |
| 12:00 PM - 12:30 PM                                   | <i>High-Throughput Functional Validation of Pathogenic Variants</i><br>Penelope Bonnen, Ph.D.<br>Baylor College of Medicine, Houston, TX                                                              |
| 12:30 PM - 12:45 PM                                   | Closing comments – END OF MEETING                                                                                                                                                                     |