

**Program for SIMD Annual Meeting
March 27-31, 2010
Hotel Albuquerque at Old Town,
Albuquerque, New Mexico**

Saturday, March 27, 2010	
3:00pm- 6:00pm	Registration for SIMD and ACMG Joint session Hotel Albuquerque – North Atrium by Alvarado Ballrooms
Sunday, March 28, 2010	
8:30am - 6:00pm	Registration for SIMD/ACMG joint session and SIMD annual meeting Hotel Albuquerque - North Atrium by Alvarado Ballrooms
12:00 noon – 6:00pm	Exhibit and poster board set up Hotel Albuquerque - Alvarado Ballrooms E,F,G,H
3:00pm	Hotel check in Hotel Albuquerque
10:00am – 3:00pm	ACMG-SIMD Joint Session: Albuquerque Convention Center Metabolic Causes of Autism and Neurodevelopmental Disability Moderators: Gerard Berry, M.D. & Nicola Longo, M.D.
9:45am	Shuttle bus to leave Hotel Albuquerque for ACMG/SIMD Joint session at Albuquerque Convention Center
10:30am – 10:40am	Opening comments – Gerard Berry, M.D.
10:40am -11:20am	Autism spectrum disorders: what they are and how deeply to test for an etiology <i>Isabelle Rapin, M.D.</i> <i>Albert Einstein College of Medicine, Bronx, NY</i>
11:20am – 12:00 noon	Metabolic alterations in autism spectrum disorders <i>Steve Kahler, M.D.</i> <i>Arkansas Children's Hospital, Little Rock, AR</i>
12:00 noon – 1:00pm	Lunch on your own
1:00pm – 1:40pm	Mitochondrial disorders and developmental delays <i>Salvatore DiMauro, MD.</i> <i>Columbia College of Physicians and Surgeons, New York, NY</i>
1:40pm – 2:20pm	Organelle dysfunction and mental retardation <i>Nancy Braverman, M.S., M.D.</i> <i>McGill University, Montreal, Quebec</i>
2:20pm – 3:00pm	The metabolic evaluation of patients with developmental delays <i>Carol Greene, M.D.</i> <i>University of Maryland School of Medicine, Washington, DC</i>
3:20pm	Shuttle bus back to Hotel Albuquerque
7:00pm – 10:00pm	Welcome Reception at Hotel Albuquerque <i>Heavy hors D'oeuvres served</i> Alvarado Ballrooms, A,B,C,D

Monday, March 29, 2010	
7:00 am - 8:15am	Breakfast— Hotel Albuquerque—Franciscan room
8:00am -5:00pm	Registration— Hotel Albuquerque— North Atrium by Alvarado Ballrooms
Scientific Session 1 8:00am -10:15pm	Session 1. Disorders of glycosylation Moderator: Shawn McCandless, M. Hotel Albuquerque, Alvarado Ballrooms A,B,C,D
8:00am - 8:45am	How proteins are glycosylated <i>Hudson H. Freeze Ph.D.</i> <i>Burnham Institute for Medical Research, La Jolla CA</i>
8:45am - 9:30am	Congenital disorders of glycosylation <i>Fernando Scaglia, M.D., FACMG</i> <i>Baylor College of Medicine, Houston, TX</i>
9:30am - 10:15am	Muscular dystrophies, brain development, and glycosylation <i>Jose Abdenur, M.D.</i> <i>Children's Hospital of Orange California, Orange, CA</i>
10:15 am -10:45am	AM Refreshment break/Exhibits open - Alvarado Ballrooms E,F,G,H
Scientific Session 2 10:45am - 12:15pm	Session 2. Ask the Experts: Challenge the Panel with Unusual and Unknown Cases. Attendees may submit candidate cases in advance or at the meeting. Panel Experts: Gerard Berry, M.D. and Steve Kahler, M.D Hotel Albuquerque - Alvarado Ballrooms A,B,C,D
12:15 pm -1:00pm	Box lunch served in Franciscan room
1:00pm -1:45pm	Robert Guthrie Memorial Lecture Population based newborn screening for Severe Combined immunodeficiency <i>Jennifer Puck, MD</i> <i>University of California, San Francisco</i> Hotel Albuquerque, Alvarado Ballrooms A,B,C,D
Scientific Session 3 2:00pm - 6:00pm	Session 3: Contributed Papers —Presentations from Travel Award Winners Moderators: Georgianne Arnold MD and Katrina Dipple, M.D. 12 presentations - 15 min each Hotel Albuquerque - Alvarado Ballrooms A,B,C,D
2:00pm - 2:15pm	The role of anti-rhGAA antibody titers and clinical Outcomes in infantile pompe disease patients <i>Suhrad Banugaria, M.B.B.S.</i> <i>Duke University Medical Center, Durham, NC</i>
2:15pm - 2:30pm	A mouse model of N-acetylglutamate synthase deficiency <i>Juan Cabrera-Luque, B.S.</i> <i>Children's National Medical Center, Washington, DC</i>
2:30pm - 2:45pm	Rescue and long-term outcome of a murine model of methylmalonic acidemia following liver-directed, AAV-mediated gene therapy <i>Nuria Carrillo-Carrasco, M.D.</i> <i>National Human Genome Research Institute/NIH, Bethesda, MD</i>
2:45pm - 3:00pm	Proteomics reveals new insights into the cause of hyperammonemia in methylmalonic acidemia <i>Randy Chandler, Ph.D.</i> <i>National Human Genome Research Institute/NIH, Bethesda, MD</i>
3:00pm - 3:15pm	Measurement of N-acetylglactosaminidase in mucopolipidosis II and III. Evidence for alternative targeting to the lysosome? <i>Irene DeBiase, M.D.</i> <i>Greenwood Genetic Center, Greenwood, SC</i>
3:15pm - 3:30pm	Relevance of dietary behaviors in phenylketonuria patients when evaluating BH4 (tetrahydrobiopterin) treatment response <i>Teresa Douglas MS</i>

	<i>Emory University, Decatur, GA</i>
3:30pm – 3:45pm	The functional phenotype of an inborn error of metabolism: outlining impairment and disability in methylmalonic acidemia <i>Natalie Hauser, M.D.</i> <i>National Human Genome Research Institute/NIH, Bethesda, MD</i>
3:45pm - 4:15pm	PM Refreshment break/Exhibits open - Alvarado Ballrooms E,F,G,H
4:15pm – 4:30pm	Compensation of deficient fatty acid oxidation in long chain Acyl-CoA dehydrogenase knockout mice <i>Sander M. Houten, Ph.D.</i> <i>Academic Medical Center, Amsterdam, The Netherlands</i>
4:30pm – 4:45pm	Outcome of propionic acidemia treated at presentation with N-Carbamylglutamate <i>Sebastien Levesque, M.D.</i> <i>Montreal Children's Hospital, McGill University, Montreal, Quebec</i>
4:45pm – 5:00pm	Desmosterolosis: Clinical presentation, neuroradiologic findings, and potential therapeutic considerations <i>Christian Schaaf, M.D.</i> <i>Baylor College of Medicine, Houston, TX</i>
5:00pm – 5:15pm	The biochemical classification of 50 <i>ACADM</i> variants obtained from five years of full gene sequencing <i>Emily Smith, Ph.D.</i> <i>Mayo Clinic, Rochester, MN</i>
5:15pm – 5:30pm	A formiminoglutamic aciduria patient found by newborn screening and the association of FTCD genotype with clinical phenotype <i>Wei Zhang, M.D.</i> <i>Emory University, Decatur, GA</i>
5:30pm – 5:45pm	Presentation from the 2009 Emmanuel Shapira Award Winner <i>Georgianne Arnold, M.D.</i> <i>A Delphi Clinical Practice Protocol for the management of very long chain Acyl-CoA dehydrogenase deficiency</i>
5:45pm – 6:00pm	Presentation from 2009 Hyperion Fellow Arginine flux and nitric oxide production in subjects with MELAS syndrome and the effect of arginine and citrulline supplementation: Study design <i>Ayman El-Hattab, M.D.</i> <i>Baylor College of Medicine, Houston, TX</i>
	Evening free in Albuquerque

Tuesday, March 30, 2010

7:00am – 8:15am	AM Breakfast— Hotel Albuquerque - Franciscan room
8:00am –12:00 Noon	Registration - Hotel Albuquerque— North Atrium by Alvarado Ballrooms
Scientific Session 4 8:00am –11:45am	Session 4. Pediatric Neurotransmitter Disorders Moderator: Nicola Longo, MD, PhD Hotel Albuquerque - Alvarado Ballrooms A,B,C,D
8:00am - 8:45am	Disorders of tetrahydrobiopterin metabolism presenting with and without hyperphenylalaninemia Nenad Blau Ph.D. <i>University Children's Hospital, Zurich, Switzerland</i>
8:45am – 9:15am	Glycine encephalopathy and disorders of serine metabolism. <i>Johan Van Hove M.D., Ph.D.</i> <i>University of Colorado at Denver, Aurora, CO</i>
9:15am – 9:45am	Pyridoxine-responsive seizures. Renata Gallagher, M.D., Ph.D. <i>The Children's Hospital, Aurora, CO</i>

9:45am- 10:15am	Heritable disorders of GABA (4-aminobutyrate) metabolism. An update <i>K. Michael Gibson, Ph.D.</i> <i>Michigan Technological University, Houghton, MI</i>
10:15am - 10:45am	AM Refreshment Break/Exhibits open - Alvarado Ballrooms E,F,G,H
10:45am - 11:15am	Tyrosine hydroxylase, amino acid decarboxylase deficiency and DOPA-responsive dystonia Kathy Swoboda, M.D. <i>University of Utah School of Medicine, Salt Lake City, UT</i>
11:15am - 11:45am	Laboratory testing for neurotransmitter disorders, <i>Keith Hyland, Ph.D.</i> <i>Medical Neurogenetics, Atlanta, GA</i>
11:45am	Adjourn - Afternoon free to enjoy Albuquerque
12:00 noon - 2:00pm	SIMD Board meeting -Potter's Room
1:00pm - 2:30pm	Optional-NIH officials will talk about current funding opportunities Hotel Albuquerque - Alvarado Ballroom, A,B,C,D
1:00pm - 5:00pm	Posters and exhibits on display - Alvarado Ballrooms E,F,G,H
3:30pm - 6:00pm	Posters Attended by Authors and Exhibits open Wine and cheese served Hotel Albuquerque - Alvarado Ballrooms E,F,G,H
6:00pm - 7:15pm	Optional Dinner (Ticket required) - Hotel Albuquerque—Franciscan room
7:30pm -8:00pm	Donough O'Brien Presidential Address The contribution of heterozygosity to disease <i>Georgirene Vladutiu, Ph.D.</i> <i>The State University of New York at Buffalo, Buffalo, NY</i> Hotel Albuquerque—Alvarado Ballrooms A,B,C,D
8:00pm - 9:30pm	SIMD Business Meeting and Award Presentations Emmanuel Shapira SIMD Award (First Author of Best Publication in MGM) Neil Buist Award (Best Oral Presentation by a Trainee) Hotel Albuquerque—Alvarado Ballrooms A,B,C,D
Wednesday, March 31, 2010	
7:00am - 8:15am	Breakfast— Hotel Albuquerque at Old Town —Franciscan room
Scientific Session 5 8:00am -10:15am	Session 5. Current treatment and outcome of Inborn Errors of Metabolism Moderator: Rani Singh, Ph.D., R.D. Hotel Albuquerque - Alvarado Ballrooms A,B,C,D
8:00am - 8:15am	Introduction: Developing nutritional guidelines for metabolic disorders <i>Rani Singh, Ph.D., R.D.</i> <i>Emory University, Decatur, GA</i>
8:15am - 8:45am	Dietary guidelines for Maple Syrup Urine Disease <i>Dianne M. Frazier PhD MPH RD</i> <i>University of North Carolina, Chapel Hill, NC</i>
8:45am - 9:15am	Dietary guidelines for Propionic acidemia <i>Keiko Ueda, MPH, RD, LDN</i> <i>Floating Hospital for Children at Tufts Medical Center, Boston, MA</i>
9:15am - 9:45am	Essential fatty acids in phenylketonuria <i>Sarah Yi, MS</i> <i>Emory University, Decatur, Georgia.</i>
9:45am - 10:15am	Novel therapies for phenylketonuria <i>Cary Harding. M.D.</i> <i>Oregon Health & Science University, Portland, OR</i>
10:15am -10:45am	AM Refreshment Break/Exhibits open - Alvarado Ballrooms E,F,G,H
Scientific Session 6 10:45am - 11:45am	Session 6. Hearing and vision loss in metabolic disorders Moderator: Bruce Barshop, M.D.

	Hotel Albuquerque—Alvarado Ballrooms A,B,C,D
10:45am - 11:15am	Pathogenic mechanisms in the neuronal ceroid- lipofuscinoses <i>Michael Bennett, Ph.D.</i> <i>The Children's Hospital of Philadelphia, Philadelphia, PA</i>
11:15am - 11:45am	Mitochondrial disorders and hearing loss <i>Doug Wallace, M.D.</i> <i>University of California at Irvine, Irvine, CA</i>
11:45am - 12:00 noon	Closing comments and meeting adjourns <i>Nicola Longo, M.D.</i> <i>University of Utah, Salt Lake City, UT</i>