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

## Saturday, March 27, 2010

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
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| 3:00pm– 6:00pm| Registration for SIMD and ACMG Joint session  
Hotel Albuquerque – North Atrium by Alvarado Ballrooms |

## Sunday, March 28, 2010

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
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| 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  
*Isabelle Rapin, M.D.*  
*Albert Einstein College of Medicine, Bronx, NY* |
| 11:20am – 12:00 noon| Metabolic alterations in autism spectrum disorders  
*Steve Kahler, M.D.*  
*Arkansas Children's Hospital, Little Rock, AR* |
| 12:00 noon – 1:00pm| Lunch on your own |
| 1:00pm – 1:40pm| Mitochondrial disorders and developmental delays  
*Salvatore DiMauro, MD.*  
*Columbia College of Physicians and Surgeons, New York, NY* |
| 1:40pm – 2:20pm| Organelle dysfunction and mental retardation  
*Nancy Braverman, M.S., M.D.*  
*McGill University, Montreal, Quebec* |
| 2:20pm – 3:00pm| The metabolic evaluation of patients with developmental delays  
*Carol Greene, M.D.*  
*University of Maryland School of Medicine, Washington, DC* |
| 3:20pm         | Shuttle bus back to Hotel Albuquerque |
| 7:00pm – 10:00pm| Welcome Reception at Hotel Albuquerque  
*Heavy hors D’oeuvres served*  
Alvarado Ballrooms, A,B,C,D |
### Scientific Session 1
8:00am - 10:15am

**Session 1. Disorders of glycosylation**

**Moderator: Shawn McCandless, M.D.**
Hotel Albuquerque, Alvarado Ballrooms A,B,C,D

<table>
<thead>
<tr>
<th>Time</th>
<th>Topic</th>
<th>Speaker</th>
<th>Affiliation</th>
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</thead>
<tbody>
<tr>
<td>8:00am</td>
<td>How proteins are glycosylated</td>
<td>Hudson H. Freeze, Ph.D.</td>
<td>Burnham Institute for Medical Research, La Jolla, CA</td>
</tr>
<tr>
<td>8:45am</td>
<td>Congenital disorders of glycosylation</td>
<td>Fernando Scaglia, M.D., FACMG</td>
<td>Baylor College of Medicine, Houston, TX</td>
</tr>
<tr>
<td>9:30am</td>
<td>Muscular dystrophies, brain development, and glycosylation</td>
<td>Jose Abdenur, M.D.</td>
<td>Children's Hospital of Orange California, Orange, CA</td>
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</tbody>
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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

### Scientific Session 3
2:00pm – 6:00pm

**Session 3: Contributed Papers**—Presentations from Travel Award Winners

**Moderators: Georjianne Arnold, MD and Katrina Dipple, M.D.**

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<tr>
<th>Time</th>
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<tbody>
<tr>
<td>2:00pm</td>
<td>The role of anti-rhGAA antibody titers and clinical Outcomes in infantile Pompe disease patients</td>
<td>Suhrad Banugaria, M.B.B.S.</td>
<td>Duke University Medical Center, Durham, NC</td>
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<tr>
<td>2:15pm</td>
<td>A mouse model of N-acetylglutamate synthase deficiency</td>
<td>Juan Cabrera-Luque, B.S.</td>
<td>Children's National Medical Center, Washington, DC</td>
</tr>
<tr>
<td>2:30pm</td>
<td>Rescue and long-term outcome of a murine model of methylmalonic acidemia following liver-directed, AAV-mediated gene therapy</td>
<td>Nuria Carrillo-Carrasco, M.D.</td>
<td>National Human Genome Research Institute/NIH, Bethesda, MD</td>
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<tr>
<td>2:45pm</td>
<td>Proteomics reveals new insights into the cause of hyperammonemia in methylmalonic acidemia</td>
<td>Randy Chandler, Ph.D.</td>
<td>National Human Genome Research Institute/NIH, Bethesda, MD</td>
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<tr>
<td>3:00pm</td>
<td>Measurement of N-acetylagalactosaminidase in mucolipidosis II and III. Evidence for alternative targeting to the lysosome?</td>
<td>Irene DeBiase, M.D.</td>
<td>Greenwood Genetic Center, Greenwood, SC</td>
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<tr>
<td>3:15pm</td>
<td>Relevance of dietary behaviors in phenylketonuria patients when evaluating BH4 (tetrahydrobiopterin) treatment response</td>
<td>Teresa Douglas</td>
<td>MS</td>
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<td>Time</td>
<td>Presentation</td>
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| 3:30pm – 3:45pm | The functional phenotype of an inborn error of metabolism: outlining impairment and disability in methylmalonic acidemia  
Natalie Hauser, M.D.  
National Human Genome Research Institute/NIH, Bethesda, MD |
| 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  
Sander M. Houten, Ph.D.  
Academic Medical Center, Amsterdam, The Netherlands |
| 4:30pm – 4:45pm | Outcome of propionic acidemia treated at presentation with N-Carbamylglutamate  
Sebastian Levesque, M.D.  
Montreal Children’s Hospital, McGill University, Montreal, Quebec |
| 4:45pm – 5:00pm | Desmosterolosis: Clinical presentation, neuroradiologic findings, and potential therapeutic considerations  
Christian Schaaf, M.D.  
Baylor College of Medicine, Houston, TX |
| 5:00pm – 5:15pm | The biochemical classification of 50 ACADM variants obtained from five years of full gene sequencing  
Emily Smith, Ph.D.  
Mayo Clinic, Rochester, MN |
| 5:15pm – 5:30pm | A formiminoglutamic aciduria patient found by newborn screening and the association of FTCD genotype with clinical phenotype  
Wei Zhang, M.D.  
Emory University, Decatur, GA |
| 5:30pm – 5:45pm | Presentation from the 2009 Emmanuel Shapira Award Winner  
Georgianne Arnold, M.D.  
A Delphi Clinical Practice Protocol for the management of very long chain Acyl-CoA dehydrogenase deficiency |
| 5:45pm – 6:00pm | Presentation from 2009 Hyperion Fellow  
Ayman El-Hattab, M.D.  
Baylor College of Medicine, Houston, TX |

**Tuesday, March 30, 2010**

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<tr>
<th>Time</th>
<th>Event</th>
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<tr>
<td>7:00am – 8:15am</td>
<td>AM Breakfast— Hotel Albuquerque - Franciscan room</td>
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<tr>
<td>8:00am –12:00 Noon</td>
<td>Registration - Hotel Albuquerque— North Atrium by Alvarado Ballrooms</td>
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| **Scientific Session 4**  
8:00am –11:45am | **Session 4. Pediatric Neurotransmitter Disorders**                  |
| 8:00am - 8:45am | Disorders of tetrahydrobiopterin metabolism presenting with and without hyperphenylalaninemia  
Nenad Blau Ph.D.  
University Children’s Hospital, Zurich, Switzerland |
| 8:45am – 9:15am | Glycine encephalopathy and disorders of serine metabolism.  
Johan Van Hove M.D., Ph.D.  
University of Colorado at Denver, Aurora, CO |
| 9:15am – 9:45am | **Pyridoxine-responsive seizures.**  
Renata Gallagher, M.D., Ph.D.  
The Children’s Hospital, Aurora, CO |
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<tr>
<td>9:45am – 10:15am</td>
<td>Heritable disorders of GABA (4-aminobutyrate) metabolism. An update</td>
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<td>K. Michael Gibson, Ph.D.</td>
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<td>Michigan Technological University, Houghton, MI</td>
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<td>10:15am – 10:45am</td>
<td>AM Refreshment Break/Exhibits open – Alvarado Ballrooms E,F,G,H</td>
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<tr>
<td>10:45am – 11:15am</td>
<td>Tyrosine hydroxylase, amino acid decarboxylase deficiency and DOPA-responsive dystonia</td>
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<td>Kathy Swoboda, M.D.</td>
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<td>University of Utah School of Medicine, Salt Lake City, UT</td>
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<tr>
<td>11:15am – 11:45am</td>
<td>Laboratory testing for neurotransmitter disorders,</td>
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<td>Keith Hyland, Ph.D.</td>
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<td>Medical Neurogenetics, Atlanta, GA</td>
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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

6:00pm - 7:15pm Optional Dinner (Ticket required) - Hotel Albuquerque—Franciscan room

7:30pm –8:00pm Donough O’Brien Presidential Address

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

Rani Singh, Ph.D., R.D.

Emory University, Decatur, GA

8:15am – 8:45am Dietary guidelines for Maple Syrup Urine Disease

Dianne M. Frazier PhD MPH RD

University of North Carolina, Chapel Hill, NC

8:45am – 9:15am Dietary guidelines for Propionic acidaemia

Keiko Ueda, MPH, RD, LDN

Floating Hospital for Children at Tufts Medical Center, Boston, MA

9:15am – 9:45am Essential fatty acids in phenylketonuria

Sarah Yi, MS

Emory University, Decatur, Georgia.

9:45am – 10:15am Novel therapies for phenylketonuria

Cary Harding, M.D.

Oregon Health & Science University, Portland, OR

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.
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<thead>
<tr>
<th>Time</th>
<th>Session Title</th>
<th>Speaker(s)</th>
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<tbody>
<tr>
<td>10:45am – 11:15am</td>
<td>Pathogenic mechanisms in the neuronal ceroid- lipofuscinoses</td>
<td>Michael Bennett, Ph.D. &lt;br&gt; <em>The Children’s Hospital of Philadelphia, Philadelphia, PA</em></td>
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<tr>
<td>11:15am – 11:45am</td>
<td>Mitochondrial disorders and hearing loss</td>
<td>Doug Wallace, M.D. &lt;br&gt; <em>University of California at Irvine, Irvine, CA</em></td>
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<tr>
<td>11:45am – 12:00 noon</td>
<td>Closing comments and meeting adjourns</td>
<td>Nicola Longo, M.D. &lt;br&gt; <em>University of Utah, Salt Lake City, UT</em></td>
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