

**SOUTHEAST NEWBORN SCREENING AND GENETICS COLLABORATIVE (SERC)  
AND  
SOUTHEASTERN REGIONAL GENETICS GROUP, Inc™ 28<sup>th</sup> ANNUAL MEETING (SERGG)**

July 22-24, 2010  
Sawgrass Marriott Hotel  
Ponte Vedra Beach, FL

Hosted by Nemours Children's Clinic

This meeting is supported in part by a grant from the Maternal and Child Health Bureau (Title V, Social Security Act), Grant #2-U22 MC010979, Health Resources and Services Administration, Department of Health and Human Services.

**Thursday, July 22, 2010 (SERC)**

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|--------------------------|--|
| 8:00 am – 10:00 am       | SERC Lab Performance Workgroup<br>David Ledbetter, PhD, Chair  |
| 10:00 am – 12:00 pm      | SERC Emergency Preparedness Workgroup<br>Hans Andersson, MD, Chair, Bill Perry, Consultant<br>Tabletop Exercise  |
| 12:00 pm – 1:30 pm       | SERGG Lysosomal Workgroup Meeting  |
| 12:00 pm – 5:00 pm       | Registration –   |
| <b>1:30 pm – 6:30 pm</b> | <b>Update on the HRSA Grant Initiative: Report on New Grant Goals and Opportunities in SERC</b>  |
| 1:30 pm – 1:45 pm        | Introduction<br>Rani Singh, PhD, RD, Project PI  |
| 1:45 pm – 2:15 pm        | HRSA/MCHB Update<br>Sara Copeland, MD, Medical Officer Genetic Services Branch   |
| 2:15 pm – 2:30 pm        | NCC Update   |
| 2:30 pm – 2:45 pm        | Medical Home<br>Alex Kemper, MD, Workgroup Chair   |
| 2:45 pm – 3:00 pm        | Sickle Cell Workgroup<br>Jim Eckman, MD, Workgroup Chair   |
| 3:00 pm – 3:15 pm        | Transitioning Medical Care into Adulthood  |
| 3:15 pm – 4:30 pm        | Laboratory Performance<br>Tim Wood, PhD  |
| 3:15 pm – 3:30 pm        | Tyrosinemia Testing<br>Catherine Evans, PhD, LA DOH  |
| 3:30 pm – 3:45 pm        | 2 <sup>nd</sup> Tier Testing<br>Miao He, PhD, Emory University   |
| 3:45 pm – 4:00 pm        | New Tests in Region 3<br>Dan Sharer, PhD, UAB  |
| 4:00 pm – 4:30 pm        | Panel Discussion   |
| 4:30 pm – 4:45 pm        | Break  |
| 4:45 pm – 5:30 pm        | Short-Term Follow-up Panel – Tricia Page, Chair<br>Penny Edwards, MS, RD, LD – FL Children's Medical Services<br>Dianne Frazier, PhD, MPH – University of NC at Chapel Hill<br>Kathy Tomashitis, MNS, RD, LD, South Carolina DOH |
| 5:30 pm – 5:45 pm        | Long-Term Follow-up Project Update<br>Rani Singh, PhD  |
| 5:45 pm – 6:15 pm        | "The Importance of Standardization of Language for Informatic Systems"<br>Alan E. Zuckerman, MD – Georgetown University Medical Center   |
| 6:15 pm – 6:30 pm        | Wrap up and Closing Remarks  |
| 6:30 pm                  | Dinner on your own   |

**Friday, July 23, 2010 (SERGG)**

- 7:00 am – 5:00 pm Registration  
7:00 am – 8:00 am Board of Directors Breakfast Meeting  
7:00 am – 8:00 am Continental Breakfast  
7:00am – 1:30 pm Vendor Exhibits  
8:00 am – 8:15 am Welcome/Announcements  
**Host: Pamela Arn, MD, Nemours Children’s Clinic**  
8:15 am – 10:15 am Platform Session 1  
**Moderator: Pamela Arn, MD, Nemours Children’s Clinic**  
8:15 am – 9:00 am **Invited Speaker**  
**“Genetic Evaluation of Short Stature”**  
Mira B. Irons, MD  
Associate Professor of Pediatrics  
Associate Chief, Division of Genetics  
Chief, Clinical Genetics and Metabolism  
Children’s Hospital Boston  
9:00 am – 9:15 am **“The Emerging Phenotype of Long-term Pompe Survivors”**  
Sean N. Prater, MRes  
Duke University Medical Center  
9:15 am – 9:30 am **“Enzyme Replacement Therapy with Velaglucerase Alfa Significantly Improves Key Clinical Parameters in Type 1 Gaucher Disease: Positive Results from a Randomized, Double-blind, Global, Phase III Study”**  
Eric Crombez, MD  
Shire Human Genetic Therapies  
9:30 am – 9:45 am **“Alu Insertion Mutation in Duchenne Muscular Dystrophy”**  
Syed Hussain Askree, MD, PhD  
Emory University School of Medicine  
9:45 am – 10:00 am **“Molecular Analysis and Protein Processing in Late-onset Pompe Patients with Low Levels of Acid Alpha Glucosidase (GAA)”**  
Deeksha Bali, PhD  
Duke University Medical Center  
10:00 am – 10:15 am **“A Successful Tool to Predict Antibodies Against Therapeutic Proteins: Experience from Infantile Pompe Disease”**  
Suhrad G. Banugaria, MBBS  
Duke University Medical Center  
10:15 am – 10:30 am **“Utility of Serum Biotinidase Activity as a Biomarker for Evaluating and Monitoring Hepatic Glycogen Storage Disorders”**  
Adviye A. Tolun, MS, PhD  
Duke University Medical Center  
10:30 am – 11:00 am BREAK  
11:00 am – 1:30 pm Platform Session 2  
**Moderator: Bruce Korf, MD, University of Alabama at Birmingham**  
11:00 am – 11:30 am **Invited Speaker**  
**“What’s New in Newborn Screening”**  
Brad L. Therrell, PhD  
Director, Newborn Screening and Genetics Resource Center  
Professor of Pediatrics, University of Texas Health Science Center  
11:30 am – 11:45 pm **“An Evaluation of the CFTR Mutation Panel Used in Second Tier Cystic Fibrosis Newborn Screening: The First Three Years in Georgia”**  
Lora J. H. Bean, PhD  
Emory University School of Medicine  
11:45 am – 12:00 pm **“Development of Reference Dried Blood Spot Materials for QPCR Assays of T-cell Receptor Excision Circles in Newborn Screening for Severe Combined Immunodeficiency”**  
Francis K. Lee, MSc, PhD  
Centers for Disease Control

- 12:00 pm – 12:15 pm ***“Comprehensive Molecular and Biochemical Analysis of Congenital Disorders of Glycosylation”***  
Miao He, PhD  
Emory University School of Medicine
- 12:15 pm – 12:30 pm ***“Quantification of Testosterone by LC-MS/MS in Dried Blood Spots for Short-term and Long-term Follow-up of Congenital Adrenal Hyperplasia”***  
N Lin, PhD  
Emory University School of Medicine
- 12:30 pm – 1:00 pm ***“Novel Methods in Newborn Screening”***  
David Millington, PhD  
Duke University School of Medicine

- 1:00 pm – 2:00 pm Board of Directors Lunch Meeting (NBS Connect Presentation)
- 1:00 pm – 5:30 pm Free Time
- 5:30 pm – 7:30 pm **Reception and Poster Session – Exhibits Open**
- 7:30 pm – 9:30 pm Concurrent Breakout Sessions**  
Nutrition/Biochemical/Clinical  
Public Health  
Cytogenetics/Molecular Genetics

**Concurrent SERC Consumer Alliance Session**

**Friday, July 23, 2010 (SERC)**

- 9:00 am – 9:15 am Welcome and Introductions  
Mike McConnell
- 9:15 am – 9:45 am NBS Connect update  
Sarah Travis
- 9:45 am – 10:30 am MSUD-Support Meeting recap  
TBD
- 10:30 am – 11:15 am Family Panel (Consumer Representatives)  
Reports from 2009-2010 Meetings  
Activities from each state
- 11:15 am – 11:30 am Break
- 11:30 am – 1:00 pm Discussion  
Strategies for starting state groups  
Marketing/networking  
Fundraising ideas
- 1:00 pm – 1:15 pm Closing remarks  
Mike McConnell
- 1:30 pm – 2:30 pm Family lunch

**Saturday, July 24, 2010 (SERGG)**

7:00 am – 8:00 am Continental Breakfast

7:15 am – 10:30 am Vendor Exhibits

8:00 am – 10:30 am Platform Session 3

**Moderator: Lori Farmer, RN, ARNP, MS, MSN, APNG, University of North Carolina at Pembroke**

8:00 am – 8:30 am **Invited Speaker**

***“Issues in Genetic Testing in Navajo Populations”***

Murray H. Brilliant, PhD  
Director of the Center for Human Genetics  
Marshfield Clinic

8:30 am – 8:45 am ***“Georgia Newborn Screening Program: Improvements in Sickle Cell Follow-up”***

Sharon Quarry, MS  
Georgia Department of Human Resources

8:45 am – 9:00 am ***“Newborn Screening (NBS) Results Reporting: A National Snapshot in 2010”***

Phaidra Floyd-Browning, RN  
Tulane University Medical School

9:00 am – 9:15 am ***“Florida and Texas Coop Experience”***

Lois Taylor, RN, BSN, CPM  
Florida Newborn Screening Follow-up Program

9:15 am – 9:30 am ***“Initial Development of an Emergency Preparedness Plan at the Greenwood Genetic Center”***

Tim Wood, PhD  
Greenwood Genetic Center

9:30 am – 9:45 am ***“Survey Results on Insurance Coverage and Financial Barriers to Care for Children with Metabolic Disorders in the Southeast Region”***

Mary C. Brauchla, MPH  
Emory University School of Medicine

9:45 am – 10:00 am ***“Diagnosis of Sanfilippo Syndrome by Enzyme Testing in Leukocytes”***

Laura M. Pollard, PhD  
Greenwood Genetics Center

10:00 am – 10:15 am ***“Measurement of N-Acetylgalactosaminidase in Mucopolipidosis II and III. Alternative Targeting to the Lysosome?”***

Irene De Biase, MD, PhD  
Greenwood Genetics Center

10:15 am – 10:30 am ***“Recurrent Translocations Mediated by Homologous Recombination”***

Katie Rudd, PhD  
Emory University School of Medicine

10:30 am – 11:00 am Break

11:00 am – 1:30 pm Platform Session 4

**Moderator: Priya Kishnani, MD, Duke University School of Medicine**

11:00 am – 12:00 pm **Invited Speakers**

***“Cytogenetic Arrays in Clinical Practice: Developing Standards for Constitutional and Cancer Applications”***

David H. Ledbetter, PhD  
Director, Division of Medical Genetics  
Emory University School of Medicine

Marilyn Li, MD  
Associate Professor  
Tulane University Medical School

12:00 pm – 12:15 pm ***“Common Leukemia- and Lymphoma-associated Genetic Arrangements and Constitutional t(11:22) in Healthy Individuals”***

Jambo Song, PhD  
Tulane University Medical School

- 12:15 pm – 12:30 pm ***“Detection of 15q11.2-q13 Copy Number Abnormalities by Quantitative DNA Methylation Analysis”***  
Bradford Coffee, PhD  
Emory University School of Medicine
- 12:30 pm – 12:45 pm ***“Copy Number Alterations in Myelodysplastic Syndrome”***  
Xiaofeng Hu, MD, PhD  
Tulane University Medical School
- 12:45 pm – 1:00 pm ***“Further Refining the Critical Region at 22q11 Associated with Autosomal Dominant Opitz Syndrome”***  
Anna K. Allred, BS  
University of Mississippi Medical Center
- 1:00 pm – 1:15 pm ***“Carrier Screening for 90 Recessive Diseases Using a Targeted Resequencing Array”***  
Alice Tanner, PhD, MS, CGC  
Emory University School of Medicine
- 1:15 pm – 1:30 pm ***“Assessment of Solution and Highly Multiplexed PCR-based Target Enrichment Using Next Generation Sequencing for Mutation Detection for Congenital Muscular Dystrophies”***  
C. Alexander Valencia, PhD  
Emory University School of Medicine
- 1:30 pm SERGG Business Meeting and Student Awards Presentation  
Rani H. Singh, PhD, RD, SERGG President
- 2:00 pm Adjournment

**Concurrent SERC Consumer Alliance Session**

**Saturday, July 24, 2010 (SERC)**

- 9:00 am – 11:00am Medical Foods Advocacy  
9:00 am – 9:45 am – Florida (TBD)  
9:45 am – 10:30 am – National Legislation (TBD)  
10:30 am – 11:00 am – State representatives
- 11:00 am – 11:15 am Break
- 11:15 am – 11:45 am Discussion – next steps for 2011
- 11:45 am – 12:00 pm Adjournment - Mike McConnell

## **Concurrent SERC Sickle Cell Workgroup**

### **Saturday, July 24, 2010 (SERC)**

8:00 am – 8:30 am	Welcome and Introductions Jim Eckman, MD and Sharon Lewis, Co-Chairs
8:30 am – 9:30 am	RuSH Update CDC Perspective  Florida Georgia North Carolina
9:30 am – 10:30 am	LTFU/NBSTRN Sickle Specific Minimal Dataset  Discussion of Minimum dataset
10:30 am – 10:45 am	Break
10:45 am – 12:00 pm	NIH Sickle carrier Research Agenda Report from the June 3-4, 2010 NIH Meeting  Sickle Cell Carrier Detection, Education and Counseling Agenda for the 21 <sup>st</sup> Century
12:00 pm – 1:00 pm	Lunch
1:00 pm – 2:30 pm	Report from the HRSA-RTI Sickle Cell Treatment Demonstration Projects (SCDTDP) Alabama North Carolina  Report from the HRSA Newborn Screening Projects
2:30 pm – 2:45 pm	Break
2:45 pm – 5:00 pm	2010 – 2011 SERC Sickle Cell Agenda Where do we go from here?
5:00 pm	Adjournment