

As of 5/20/2025

42nd ANNUAL MEETING of the SOUTHEASTERN REGIONAL GENETICS GROUP (SERGG)

July 17-19, 2025
Asheville, North Carolina

Wednesday, July 16, 2025

7:00 pm - SERGG Board of Directors Dinner Meeting – Paparazzi Room (12th Floor)

All Sessions are open to everyone!

Thursday, July 17, 2025

9:00 am – 5:00 pm Registration Open – Salon A Foyer

10:00 am – 1:15 pm Platform Session 1 – Salon C – Hans Andersson, MD, SERGG President

10:00 am – 12:00 pm **HRSA Co-Propel Meeting**
Rani Singh, PhD, LD (Grant PI) and Serei North, MPH (Grant Program Manager)
Updates on Southeast Integrative Newborn Screening and Long-Term Follow-Up (SE INBS-LTFU)

“Breaking Down Barriers: How a Legislative Change is Improving Newborn Screening Follow-up”
Lauren Thompson, DO, Medical University of South Carolina

12:15 pm – 1:15 pm **Industry Supported Symposium – Windsor Ballroom**
“Beyond Storage: New Insights into Lysosomal Diseases and the Potential for Next Generation SRTs”
Hosted by Sanofi
(Box Lunch provided for attendees *(CEU Not Provided)*)

12:00 pm – 1:30 pm Lunch On Your Own

1:30 am – 1:45 am **Welcome/Announcements – Salon C** – Hans Andersson, MD, SERGG President

1:45 pm – 4:45 pm **Platform Session 2 – Salon C (CEU Provided)**
Moderator: Hans Andersson, MD, Tulane University

1:45 pm – 2:30 pm **Keynote Speaker:**
Philip J Brooks, PhD, Deputy Director, Div of Rare Diseases Res Innovation, NIH
“Therapeutic Platforms Rare Monogenic Disease Clinical Trials”

2:30 pm – 2:45 pm “Beyond Diagnosis: Treating Genetic Disorders with Targeted Therapies”
Kristen Lancaster, MD, Medical University of South Carolina

2:45 pm – 3:00 pm “From Molecules to Medicine: New Hope Offered by Synthetic Transfer RNA Technology”
Misty Smith, PhD, RN, University of Alabama in Huntsville:

3:00 pm – 3:15 pm “Substantial Dietary Intake Variation in PKU Highlights Implications for Clinical Trial Design”
Jessica Strosahl, PhD, RDN, LDN, Emory University

3:15 pm – 3:45 pm **BREAK – Salon A Foyer**

3:45 pm – 4:00 pm “Outcomes of Liver Transplantation in Glycogen Storage Disease Type IB”
Alicia Khazzeka, MD, Duke University

4:00 pm – 4:15 pm “Quantitative Muscle Ultrasound as a Window Into Disease Progression in Infantile-onset Pompe Disease”
Myriam Boueri, MD, Duke University

As of 5/20/2025

4 :15 pm – 4:30 pm	“Dried Blood Spot Arylsulfatase A Enzyme Analysis for Secon-tier Newborn Screening and Diagnosis of Metachromatic Leukodystrophy” Francyne Kubaski, PhD, Greenwood Genetic Center
4:30 pm – 4:45 pm	“Perspectives on Pharmacogenomics in the Fragile X Syndrome Community: Insights from a Quantitative Survey” Erin Whiting, BS, University of Alabama at Birmingham
5:00 pm – 7:00 pm	Welcome Reception and Poster Session (Cash Bar) – Salon A-B (CEU Not Provided)
7:00 pm – 9:00 pm	Screening of the Film “The Zebra & The Bear” Filmed over seven years, The Zebra & The Bear is a documentary film about how a mother’s fierce determination to save her daughter from a devastating ultra-rare disease leads her on a journey to raise millions of dollars and drive the development of a pioneering gene-therapy treatment.

Please place all phones on vibrate when in the meeting rooms.

Conference Room Internet Access:

Network: Renaissance_Conference

Password: avlbr2

Wi-fi is also available in the lobby areas and guest rooms.

Be sure to visit each exhibit during the reception, breaks and sessions you are not attending to get your card punched for the gift card drawing!

CEUs are being offered for Genetic Counselors for this meeting. In order to receive CEUs, you must select the CEU add on as part of the online conference registration (your registration is able to be edited) and will cost \$35.00. You must provide your NSGC ID during the online registration as well as the payment in order to receive CEUs. The CEU surveys are online this year at SERGG.org under the genetic counseling button. Please complete surveys no later than 8/31/25 for CEUs, or you may not be able to receive CEUs for the conference. **Platform Codes will be printed and displayed on the backs of the doors to the lecture room entrance. These codes MUST be included to prove attendance.** For any questions, or if you could not locate a platform attendance code, please find Katy Drazba during the meeting or ask the front registration desk. Katy’s e-mail is kdrazba@ggc.org for additional questions or concerns.

All Sessions are open to everyone!

Friday, July 18, 2025

- 8:00 am – 9:00 am** **Two Concurrent Industry Supported Symposiums (CEU Not Provided)**
(Continental Breakfast included for attendees)
- #1 – “Asofatase Alfa: Enzyme Replacement Therapy for HPP” – Swannanoa Room**
 Hosted by **Alexion Pharmaceuticals**
- #2 – “Bridging the Gap in GSDIa: Addressing unmet needs and standard of care” – Victoria Room**
 Hosted by **Ultragenyx**
- 8:00 am – 9:00 am** **Continental Breakfast – (for those not attending a Symposium) – Salon A-B**
- 8:00 am – 2:00 pm** **Exhibits and Posters Open – Salon A-B**
- 9:00 am – 3:00 pm** **Registration Open – Salon A Foyer**
- 9:00 am – 10:30 am** **Platform Session 3 – Salon C (CEU Provided)**
Moderator: Barbara DuPont, PhD, Greenwood Genetic Center
- 9:00 am – 9:45 am** **Invited Speaker: Gavin Arno, PhD, Associate Director of Research (Innovation), Greenwood Genetic Center**
 “Advanced Genomic Analysis of Inherited Eye Disease; Observations and Results from Large-Cohort Studies and Beyond”
- 9:45 am – 10:00 am** **“Early Diagnosis and Genotype Influence Outcomes in CBLCL Disorder: A Retrospective Review”**
 Julia Eazer, BS, University of South Florida Morsani College of Medicine
- 10:00 am – 10:15 am** **“Spinal Muscular Atrophy Newborn Screening: Parents’ Psychosocial Experiences Through Treatment and Onward”**
 Lauren Wilson, BS, Emory University
- 10:15 am – 10:30 am** **“Broadening Access to Care: Patient Engagement at a Southeastern Cardiogenetics Program”**
 Elizabeth Towles, BA, Medical University of South Carolina
- 10:30 am – 11:00 am** **Break with Exhibits and Posters – Salon A-B**
- 11:00 am – 12:30 pm** **Platform Session 4 – Salon C (CEU Provided)**
Moderator: Rani H Singh, PhD, RD, LD, Emory University
- 11:00 am – 11:45 am** **Invited Speaker: Yue Guan, ScM, PhD, CGC, Associate Professor, Rollins School of Public Health, Emory University**
 “A Multilevel Adaptation Approach to Expand a Statewide Cancer Genetic Risk Screening Program”
- 11:45 am – 12:00 pm** **“Evaluating the Impact of Tailored Cohort Messaging on Patient Participation in Population-wide Genomic Screening”**
 Lauren Carroll, MS, Medical University of South Carolina
- 12:00 pm – 12:15 pm** **“How Does Population Genomic Screening Impact Patients? Understanding the Patient Experience and Downstream Implications”**
 Sarah English, MS, Medical University of South Carolina
- 12:15 pm – 12:30 pm** **“Genetic Anthropology of the Acadian People”**
 John R Doucet, PhD, Nicholls State University
- 12:30 pm – 2:00 pm** **Lunch – Salon A-B with overflow seating in Windsor Ballroom**

CONCURRENT INDUSTRY-SUPPORTED SYMPOSIUM
(CEU Not Provided)

TIME			
2:00 pm – 3:00 pm	“Innovations in Genetic Testing: Designing and Curating for Maximized Clinical Utility” - Hosted by Ambry Genetics	“Optimizing Therapeutic Proteins through PEGylation” - Hosted by Chiesi	“Insights into the Unmet Needs in MPS II” - Hosted by Denali Therapeutics

3:15 pm – 4:45 pm Platform Session 5 – Salon C (CEU Provided)
Moderator: Hans Andersson, Tulane University

- 3:15 pm – 4:00 pm **Invited Speaker: Ben Solomon, MD, Clinical Director, NHGRI**
“Artificial Intelligence in Medical Genomics”
- 4:00 pm – 4:15 pm **“Leveraging Machine Learning and Metabolomics for Clinical Applications: Advancing the Diagnosis and Understanding of DEGS1 Deficiency”**
Eileen Barr, MS, CGC, Baylor Genetics
- 4:15 pm – 4:30 pm **“Breaking Records: Updates to Rapid Genomic Sequencing in Hospitalized Pediatric Patients”**
Alyssa Hoelscher, MS, University of South Carolina
- 4:30 pm – 4:45 pm **“Cone Health Geneconnect: A Collaborative, Community and Value-based, Equitable Population Health Genomic Screening Program”**
Chad R Halderman-Englert, MD, Cone Health

5:00 pm – Adjournment for the Day – Enjoy your evening in Asheville!

All Sessions are open to everyone!

Saturday, July 19, 2025

- | | |
|---------------------|---|
| 9:00 am – 10:45 am | Registration Open – Salon A Foyer |
| 8:00 am – 9:00 am | Two Concurrent Industry Supported Symposiums (CEU Not Provided)
(Continental Breakfast included for attendees)

#1 – “Advancements in Rett Syndrome (RTT) Care” – Victoria Room
Hosted by Acadia Pharmaceuticals

#2 – “The Neurotoxicity of PKU” – Shawn Christ, PhD – Swannanoa Room
Hosted by BioMarin Pharmaceuticals |
| 8:00 am – 9:00 am | Continental Breakfast – (for those not attending a Symposium) – Salon A-B |
| 8:00 am – 11:15 am | Exhibits with Posters – Salon A-B |
| 9:00 am – 10:30 am | Platform Session 6 – Salon C (CEU Provided)
Moderator: Dawn Laney, MS, CGC, Emory University |
| 9:00 am – 9:45 am | Invited Speaker: Marni Falk, MD, Exec Director, Mitochondrial Medical Frontier Program, Children’s Hospital of Philadelphia
<i>“Developing intelligent mitochondrial medicines: Novel therapeutic insights from preclinical models”</i> |
| 9:45 am – 10:00 am | “Gain-of-Glycosylation Variants in IDUA Represent a Mechanism of Disease Amenable to Treatment with Novel Glycosylation Inhibitors”
Richard Steet, PhD, Greenwood Genetic Center |
| 10:00 am – 10:15 am | “Prospective Characterization of Early Symptom Onset and Progression in Young Pediatric Patients with Variants in the GLA gene Across Five Years: Longitudinal Data from the Fabry MOPPet Study”
Dawn Laney, MS, CGC, Emory University |
| 10:15 am – 10:30 am | “Newborn Screening for Pompe Disease in Georgia: Results and Factors Affecting Timely Resolution of a Positive Screen”
Erin Kistenberg, BS, Emory University |
| 10:30 am – 11:00 am | Break with Exhibits and Posters – Salon A-B |
| 11:00 am – 12:30 pm | Platform Session 7 – Salon C (CEU Provided)
Moderator: Katy Drazba, MPH, MS, CGC, Greenwood Genetic Center |
| 11:00 am – 11:45 am | Invited Speaker: Allyn McConkie-Rosell, PhD, Professor of Pediatrics, Duke University
<i>“Facing the Unknown: The Ongoing Journey of Genomic Empowerment for Families of Children with Emerging Ultra-Rare Diagnoses”</i> |
| 11:45 am – 12:00 pm | “Breaking New Ground: The Introduction of Impatient Genetic Counselor Services in South Carolina”
Olivia Thompson, MS, Medical University of South Carolina |
| 12:00 pm – 12:15 pm | “Prenatal Pitfalls: The Diagnostic Gap Between Prenatal Screening and Postnatal Reality”
Emily Henderson, MS, Medical University of South Carolina |
| 12:15 pm – 12:30 pm | “The Newborn Screen: An Investigation of Stress and Coping Among Caregivers”
Emma Sass, BA, University of Miami |

As of 5/20/2025
12:30 pm – 1:00 pm

SERGG Business Meeting & Student Award Presentations – Salon C – Hans
Andersson, MD, SERGG President

1:00 pm

Adjournment – See you next year!

DRAFT