SOUTHEAST REGIONAL GENETICS NETWORK (SERN)
35th ANNUAL MEETING of the SOUTHEASTERN REGIONAL GENETICS GROUP (SERGG)
July 20-22, 2017
Asheville, North Carolina
This meeting is supported in part by a grant from the Maternal and Child Health Bureau (MCHB) (Title V, Social Security Act), Grant #UH7MC30772 Health Resources and Services Administration (HRSA), Department of Health and Human Services.

Wednesday, July 19, 2017

7:00 pm - **SERGG Board of Directors Dinner Meeting – Club Lounge (12th Floor)**

All Sessions and Workgroup Meetings are open to everyone!

Thursday, July 20, 2017

8:30 am – 10:00 am **Public Health Session: Long-term Follow-up Workgroup – Rani Singh, PhD, RD – VICTORIA ROOM**

8:30 am - 8:50 am **Welcome Remarks and Introductions** – Rani Singh, PhD, RD
8:50 am - 9:00 am **Newborn Screening (NBS Connect) Patient Registry Update** – Theresa Pringle, MPH, Yetsa Osara, MPH, PMP
9:00 am - 9:20 am **Newborn Screening Translational Research Network (NBSTRN) Update** – Joanne Adelberg, MS, Helena Dessie, NBSTRN, American College of Medical Genetics and Genomics (ACMG)
9:20 am - 9:40 am **Summary of Sickle Cell Activities in the Southeast Region** – James Eckman, MD
9:40 am - 10:00 am **Regional Discussion and Future Directions for LTFU** – Various State Representatives

10:00 am – 12:00 pm **Telegenetics Workgroup -** Hans Andersson, MD, Chair – **ALEXANDER ROOM**
This annual telegenetics meeting offers several presentations of diverse interest. Attendees need not have any experience. Plenty of time will be available for questions and discussion.

Opening Remarks and Updates of Telegenetics Activities – Hans Andersson, MD, Tulane University

Regional Vision of Telegenetics – Rani Singh, PhD, RD, Emory University School of Medicine

Kansas- Arkansas Telegenetic Session – Shobana Kubendran, MBBS, MS, CGC, Kansas University School of Medicine-Wichita

Family History Online Application – Brandon Welch, PhD, Medical University of South Carolina

Telegenetics (distance session from Florida by online teleconference) - Heather Stalker, MS, CGC and Robert Zori, MD, University of Florida

12:00 pm – 1:00 pm **Industry Supported Symposium – Sanofi Genzyme – SWANNANOA ROOM**
"Challenging Neurological Manifestations in Lysosomal Storage Disorders"
(Box Lunch Provided)

12:00 pm – 1:00 pm **Lunch On Your Own**

12:00 pm – 5:00 pm **Registration – SALON A FOYER**

1:00 pm – 6:00 pm **Platform Session 1 – SALON A**
Chair: Rani Singh, PhD, RD, Emory University School of Medicine

1:00 pm – 1:10 pm **Introduction** – Rani Singh, PhD, RD, Emory University School of Medicine

1:10 pm – 1:30 pm **HRSA Update**
Joan Scott, MS, CGC/Jill Shuger, ScM, HRSA

1:30 pm – 1:50 pm ***National Coordinating Center (NCC) and Genetic Services Branch, MCHB Update***
Alisha Keehn/ Megan Lyon, MPH, NCC, American College of Medical Genetics (ACMG)

1:50 pm – 2:10 pm ***"Vision for Telemedicine"***
Elizabeth A. Krupinski, PhD, Emory University School of Medicine

2:10 pm – 2:40 pm	***"Creating a Digital Footprint in Genetic Health"*** Shane Owens, MBA, MS, HCI, Georgia Tech Research Institute
2:40 pm – 3:20 pm	***Newborn Screening Studies in North Carolina*** Cynthia M. Powell, MD, University of North Carolina at Chapel Hill
3:20 pm – 3:50 pm	**Break – SALON A FOYER**
3:50 pm – 4:20 pm	***"Using Clinical Metabolomics to Screen for Inborn Errors of Metabolism"*** Kirk L. Pappan, Ph.D, Scientific Discovery and Application, Metabolon, Inc.
4:20 pm – 4:40 pm	***Genetic Counseling Licensure Report*** Katie Lang, MS, CGC, Northside Hospital Cancer Institute
4:40 pm – 5:10 pm	***"Newborn Screening for Pompe Disease: Lessons Learned from New York"*** Amy C. Yang, MD, Icahn School of Medicine at Mount Sinai
5:10 pm – 5:40 pm	**Keynote Address: *"State Statutes and Legislation Related to Coverage of Dietary Treatments of Heritable Disorders"*** Meg Comeau, MHA, Catalyst Center, Boston University School of Public Health
5:40 pm – 6:00 pm	***"New Gene Discovery and Other Insights from Diagnostic Evaluations in the Undiagnosed Diseases Network"*** Loren Pena, MD, Undiagnosed Disease Network, Duke University
6:30 pm – 8:30 pm	**Welcome Reception and Poster Session (Cash Bar) – SALON B-C** **Supported by all of the exhibitors.** Be sure to visit each exhibit at some time during the meeting and get your card punched for the gift card drawing!

Friday, July 21, 2017

All Sessions and Workgroup Meetings are open to everyone!

7:30 am – 3:00 pm **Registration – SALON A FOYER**

7:30 am – 8:30 am **Continental Breakfast – SALON B-C**
Supported by Sobi, Inc.

7:30 am – 2:45 pm **Vendor Exhibits and Posters – SALON B-C**

8:30 am – 8:45 am **Welcome/Announcements** – Pam Arn, MD, President, SERGG – **SALON A**

8:45 am – 10:15 pm **Platform Session 2 – SALON A**
Moderator: Monica Basehore, PhD, Fullerton Genetics Center

8:45 am – 9:30 am **Invited Speaker: Cindy L Vnencak-Jones PhD – Vanderbilt University**
"Precision Medicine for the Right Drug at the Right Dose on the First Time"

9:30 am – 9:45 am ***"The NC Nexus Project of Newborn Exome Sequencing"***
Cynthia Powell, MD – University of North Carolina at Chapel Hill

9:45 am – 10:00 am ***"Focused Exomes: A Targeted Genomic Test with a Safety Net"***
Eric Bend, PhD – Greenwood Genetic Center

10:00 am – 10:15 am ***"Susceptibility to Post-Traumatic Stress Disorder (PTSD) is Comorbid with Mitochondrial Dysfunction in Mice"***
Graeme Preston, BA – Tulane University School of Medicine

10:15 am – 10:45 am **Break with Exhibits and Posters – SALON B-C**
Supported by Horizon Pharma

10:45 am – 11:45 am **Platform Session 3 – SALON A**
Moderator: Amie Thompson, MS, Greenwood Genetic Center

10:45 am – 11:00 am ***"Copy Number Variant Analysis Enhances Molecular Diagnostic Yield of Inborn Errors of Metaboism"***
Yuan Yuan Ho, PhD – Invitae Corporation

11:00 am – 11:15 am ***"Metabolic Pathognomonics: Incorporating Disease-Specific Biochemical Data Improves Variant Interpretation for Inherited Metabolic Disorders"***
Laura Murillo, PhD – Invitae Corporation

11:15 am – 11:30 am ***"One Year Follow-up of B Vitamin and Iron Status in Patients with Phenylketonuria Responsive and Non-responsive to Sapropterin"***
Teresa Douglas, MS, PhD – Emory University School of Medicine

11:30 am – 11:45 am ***"At-Home Urine Test for Blood Phe-Level Monitoring for Phenylketonuria (PKU)"***
Robert Latour, PhD – Clemson University

12:00 pm – 1:00 pm **Lunch – WINDSOR BALLROOM AND FOYER**
Supported by Recordati Rare Diseases

1:00 pm – 2:45 pm **Platform Session 4 – SALON A**
Moderator: Pam Arn, MD, Nemours

1:00 pm – 1:45 pm **Invited Speaker: Karen Gripp, MD - Nemours**
"Objective Digital Facial Analysis: A New Tool for Clinicians and Laboratory Geneticists"

1:45 pm – 2:00 pm ***"Recognition of Neurofibromatosis Type I with Facial Dysmorphology Novel Analysis"***
Carley Cignetti, BS – University of Alabama at Birmingham

2:00 pm – 2:15 pm ***"X Chromosome Deletion Associated with Mosaicism in Two Generations of Females"***
Kristin May, PhD – Genetic Diagnostic Laboratory

2:15 pm – 2:30 pm ***"Using All of the Tools in the Diagnostic Toolbox for Lysosomal Storage Diseases"***
Laura Pollard, PhD – Greenwood Genetic Center

2:30 pm – 2:45 pm ***"ATP1A3 Gene Alterations in Patients with Neurologic Features and Abnormal Muscle Tone"***
Lauren Baggett, MS, CGC – Greenwood Genetic Center

2:45 pm – 3:00 pm **Travel to Concurrent Industry-Supported Symposium**

CONCURRENT INDUSTRY-SUPPORTED SYMPOSIUM

TIME	VICTORIA ROOM	SWANNANOA ROOM	BERKELEY ROOM
3:00 pm – 4:00 pm	*Challenges in UCD Management: An Interactive Case Discussion* – Horizon Pharma	*Expanding Utility for WES-based Testing* - GeneDX	*Neurodegeneration in Lysosomal Storage Disorders: Focus on Neuronopathic MPS II*- Shire
4:00 pm – 4:15 pm		**BREAK**	**BREAK**
4:15 pm – 5:15 pm		Pushing the Boundary of Genomics - PerkinElmer	*On the Frontline of Managing Patients with Lysosomal Diseases* - Shire

INDUSTRY SUPPORTED SYMPOSIUM AGENDAS

***Challenges in UCD Management: An Interactive Case Discussion* – VICTORIA ROOM**
Presented by: Horizon Pharma

Time	Presentation	Speaker
3:00 pm – 3:20 pm	Overview of UCDs	Rani Singh, PhD, RD Emory University
3:20 pm – 3:40 pm	Case Profile: Practical Neurocognitive Screening and Outcomes in Patients with UCDs	Neena L. Champaigne, MD Greenwood Genetic Center
3:40 pm – 3:50 pm	Panel discussion	
3:50 pm – 4:10 pm	Case Profile: Newborn Screening Challenges and the importance of Early Treatment of Patients with UCDs	Roberto Zori, MD University of Florida
4:10 pm – 4:20 pm	Panel discussion	
4:20 pm – 4:40 pm	Case Profile: Navigating the Nutritional Needs of the Patient with a UCD	Rani Singh, PhD, RD
4:40 pm – 4:50 pm	Panel discussion	
4:50 pm – 5:10 pm	UCD Dilemmas: Cases from the Audience • Provides attendees with an opportunity to bring interesting, intriguing, or challenging UCD cases to the attention of the panel of experts for discussion	Rani Singh, PhD, RD Neena L. Champaigne, MD Roberto Zori, MD
5:10 pm	Summary and Concluding Remarks	Rani Singh, PhD, RD

***Expanding Utility for WES-based Testing* – SWANNANOA ROOM**
Presented by: GeneDX

Time	Presentation	Speaker
3:00 pm – 4:00 pm	Expanding Utility for WES-based Testing	Elizabeth Williams, MS, CGC

***Neurodegeneration in Lysosomal Storage Disorders: Focus on Neuronopathic MPS II*– BERKELEY ROOM**
Presented by: Shire

Time	Presentation	Speaker
3:00 pm – 4:00 pm	Neurodegeneration in Lysosomal Storage Disorders: Focus on Neuronopathic MPS II	Kelly King, PhD, LP, ABPP-CN, Clinical Neuropsychologist, University of Minnesota

4:00 pm – 4:15 pm BREAK

***Next Generation Sequencing in Newborn Screening* – SWANNANOA ROOM**
Presented by: PerkinElmer

Time	Presentation	Speaker
4:15 pm – 5:15 pm	Pushing the Boundary of Genomics	Madhuri Hegde, PhD PerkinElmer

***On the Frontline of Managing Patients with Lysosomal Diseases* – BERKELEY ROOM**
Presented by: Shire

Time	Presentation	Speaker
4:15 pm – 5:15 pm	On the Frontline of Managing Patients with Lysosomal Diseases	Dawn Laney, MS, CGC Emory University

5:30 pm **Adjournment for the Day** – Enjoy your evening in Asheville!

Concurrent Consumer Alliance Session – ALEXANDER ROOM

Medical Advisory Chair: Jim Eckman, MD

8:30 am – 8:45 am	**Welcome and Introductions** – Melissa Perez, Consumer Alliance Chair
8:45 am – 9:00 am	**HRSA Grant Update, Future of Telemedicine, SERN** – Rani Singh, PhD, RD
9:00 am – 9:15 am	**Consumer Delphi Study Update** – Mary Lauren Salvatore, MPH, CHES
9:15 am – 9:45 am	**At Home Urine Test for Blood Phe-Level Monitoring for Phenylketonuria (PKU) –** Robert Latour, PhD
9:45 am – 10:15 am	**State of the State Consumer and Advocacy Updates** – Consumer Alliance
10:15 am – 10:45 am	**Break**
10:45 am – 12:00 pm	**Know Who You Are** – Communication Strategies Part 1 – Melissa Perez
12:00 pm – 1:00 pm	**Lunch – WINSOR BALLROOM AND FOYER**
1:00 pm – 2:00 pm	**Patient Perspectives Across the Metabolic Community** – Yarbrough (GA), Holland (LA), Starnes (FL), Beasley (NC), Bartke(IL) Moderated by Rani Singh, PhD, RD
2:00 pm – 2:15 pm	**Break**
2:15 pm – 3:00 pm	**Maternal PKU for NBS Connect –** Amy Cunningham, MS, LDN, RD; Dianne Frazier, PhD, MPH, RD
3:00 pm – 5:30 pm	**Join the SERGG Industry Sponsored Symposiums**

Saturday, July 22, 2017

All Sessions and Workgroup Meetings are open to everyone!

7:00 am – 8:00 am **Continental Breakfast – SALON B-C**
Supported by BioMarin

7:00 am – 10:30 am **Vendor Exhibits and Posters – SALON B-C**

8:00 am – 10:30 am **Registration**

8:00 am – 8:15 am **Announcements** – Pam Arn, MD, President, SERGG – **SALON A**

8:15 am – 9:45 am **Platform Session 5 – SALON A**
Moderator: Dan Sharer, PhD, University of Alabama at Birmingham

8:15 am – 9:00 am **Invited Speaker: Robert A. Kesterson, PhD - UAB**
"CRISPRs & Gene Editing: Laboratory Use & Clinical Future"

9:00 am – 9:15 am ***"A Neuropsychologic Model of MELAS Disease Using Inducible Pluripotent Stem Cell Derived Excitatory Neurons"***
Tamas Kozicz, MD, PhD – Tulane University School of Medicine

9:15 am – 9:30 am ***"The Heart of the Matter: Cardiac Involvement in Patients with Late-onset Pompe Disease and the C.-32-13T>G Variant"***
Mrudu Herbert, MD, PhD – Duke University School of Medicine

9:30 am – 9:45 am ***"Oral D-Galactose Supplementation in PGM1-CDG"***
Eva Morava-Kozicz, MD, PhD – Tulane University School of Medicine

9:45 am – 10:15 am **Break with Exhibits and Posters – SALON B-C**
Supported by Shire

10:15 am – 11:45 am **Platform Session 6 – SALON A**
Moderator: Art Hagar, PhD, Georgia Department of Public Health

10:15 am – 11:00 am **Invited Speaker: Emily Lisi, MS, CGC – Mayo Clinic**
"The Current Status of Direct-to-Consumer Genetic Testing"

11:00 am – 11:15 am ***"Implementation of Newborn Screening for Pompe Disease, MPS-1 and X-Linked Adrenoleukodystrophy in Georgia"***
Dawn Laney, MS, CGC, CCRC – Emory University School of Medicine

11:15 am – 11:30 am ***"Greenwood Genetic Center Telehealth Initiative"***
Michael Lyons, MD – Greenwood Genetic Center

11:30 am – 11:45 am ***"Urinary Gb_3 is a Useful Biomarker for Monitoring Treatment in Fabry Disease"***
Sarah Young, PhD – Duke University Medical Center

12:00 pm – 12:30 pm **SERGG Business Meeting and Student Award Presentations –** Pam Arn, MD, President, SERGG

12:30 pm **Adjournment** – See you next year!

Concurrent Consumer Alliance Session – SWANNANOA ROOM

Medical Advisory Chair – Jim Eckman, MD

8:00 am – 8:15 am	**Welcome –** Melissa Perez, Consumer Alliance Chair
8:15 am – 8:35 am	**Resources for Caregiver Support –** Linda Starnes
8:35 am – 9:35 am	**Communication Strategies Part 2** – Melissa Perez
9:35 am – 10:00 am	**Role of the Genetic Counselor in IEM Care –** Emily Lisi, MS, CGC
10:00 am – 10:15 am	**Break**
10:15 am – 10:35 am	**Baby's First Test Update –** Natasha Bonhomme
10:35 am – 10:55 am	**Emergency Preparedness Document Launch** – Julie Tucker
10:55 am – 11:10 am	**Introduction of New Consumer Alliance Leadership** – Melissa Perez, Frances Beasley
11:10 am – 11:45 am	**Planning for 2017-2018** – Mary Lauren Salvatore, MPH, CHES
11:45 am – 12:00 pm	**Closing Remarks** – Melissa Perez, Frances Beasley
12:00 pm – 12:30 pm	**Join SERGG Business Meeting and Awards**

SAVE THE DATE!

Future

SERN/SERGG ANNUAL MEETING

July 19-21, 2018

Renaissance Asheville Marriott Hotel

Asheville, North Carolina