MEETING LOCATIONS

DATE/TIME	FUNCTION	LOCATION
Wednesday, July 18		
6:00 p – 8:30 p	Registration	Salon A Foyer
7:00 p -	SERGG Board of Directors Dinner Meeting	Top of the Plaza
Thursday, July 19		
8:30 a – 10:00 a	Breakfast for SERN Invited Speakers	Victoria
9:00 a – 10:00 a	SERN Consumer Alliance	Alexander
10:00 a – 12:00 p	Telegenetics Workgroup	Swannanoa
12:00 p – 5:00 p	Registration	Salon A Foyer
12:15 p – 1:15 p	ISS-Sanofi Genzyme	Windsor Ballroom
1:30 p – 6:30 p	Platform Session 1	Salon A
3:30 p – 4:00 p	Break	Salon A Foyer
6:30 p – 8:30 p	Reception and Poster Session	Salon B-C
Friday, July 20		
7:00 a – 8:00 a	ISS-Shire (with breakfast)	Windsor Ballroom
7:00 a – 8:30 a	Continental Breakfast	Salon B-C
7:30 a – 2:45 p	Exhibits/Posters	Salon B-C
7:30 a – 3:00 p	Registration	Salon A Foyer
8:30 a – 10:15 a	Platform Session 2	Salon A
10:15 a – 10:45 a	Break	Salon B-C
10:45 a – 11:45 a	Platform Session 3	Salon A
12:00 p – 1:00 p	Lunch	Windsor Ballroom and Foyer
1:00 p – 2:45 p	Platform Session 4	Salon A
2:45 p – 3:00 p	Travel to Concurrent Industry Supported Symposia	
3:00 p – 5:00 p	ISS-BioMarin	Swannanoa
3:00 p – 4:00 p	ISS-Shire	Victoria
3:00 p – 4:00 p	ISS-FDNA	Windsor Ballroom
Friday, July 20 - CONCURRENT SESSION – SERN CONSUMER ALLIANCE		
8:00 a – 10:00 a	Consumer Alliance Plenary	Alexander
10:00 a – 10:15 a	Break	Alexander
10:15 a – 12:00 p	Consumer Alliance Plenary	Alexander
12:00 p – 1:00 p	Lunch, State Updates, and travel to Salon A	Alexander
1:00 p – 1:45 p	Join SERGG Platform Session 4	Salon A
1:45 p – 2:00 p	Travel to Alexander	
2:00 p – 3:15 p	Consumer Alliance Plenary	Alexander
3:15 p – 3:30 p	Closing Remarks	Alexander
Saturday, July 21		
7:00 a – 8:00 a	Continental Breakfast	Salon B-C
7:00 a – 10:30 a	Exhibits and Posters	Salon B-C
8:00 a – 10:30 a	Registration Desk Open	Salon A Foyer
8:00 a – 9:45 a	Platform Session 5	Salon A
9:45 a – 10:15 a	Break	Salon B-C
10:15 a – 11:15 a	Platform Session 6	Salon A
11:15 a – 11:45 a	SERGG Business Meeting	Salon A

SOUTHEAST REGIONAL GENETICS NETWORK (SERN)
36th ANNUAL MEETING of the SOUTHEASTERN REGIONAL GENETICS GROUP (SERGG)
July 19-21, 2018
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 18, 2018

6:00 pm – 8:30 pm **Registration – SALON A FOYER**

7:00 pm - **SERGG Board of Directors Dinner Meeting – TOP OF THE PLAZA**

All Sessions and Workgroup Meetings are open to everyone!

Thursday, July 19, 2018

8:30 am – 10:00 am **Breakfast for SERN Speakers** (by invitation) – **VICTORIA ROOM**

9:00 am – 10:00 am **SERN Consumer Alliance - ALEXANDER ROOM**
"Think Genetics"
Dawn Laney, MS, CGC, CCRC, Emory University School of Medicine

10:00 am – 12:00 pm **Telegenetics Workgroup -** Hans Andersson, MD, Chair – **SWANNANOA 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

Telegenetics Clinic Demonstration – Michael J. Lyons, MD, and Katy Drazba, MPH, MS, CGC, Greenwood Genetics Center
Discussion

TRC training modules – Lloyd Sirmons, Southeast Telehealth Resource Center

"Using ItRunsInMyFamily to Share Genetic Test Results with Relatives"
Brandon Welch, PhD, Medical University of South Carolina

12:15 pm – 1:15 pm **Industry Supported Symposium – Sanofi Genzyme – WINDSOR BALLROOM**
"Cardiovascular Complications in Lysosomal Storage Diseases"
(Box Lunch provided for attendees)

12:00 pm – 1:30 pm **Lunch On Your Own**

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

1:30 pm – 6:30 pm **Platform Session 1 – SALON A**
Chair: Rani Singh, PhD, RD, Emory University School of Medicine

1:30 pm – 1:40 pm **Introduction** – Rani Singh, PhD, RD, Emory University School of Medicine

1:40 pm – 2:00 pm **HRSA Update**
Debi Sarkar, MPH/Jill Shuger, ScM, HRSA

2:00 pm – 2:20 pm ***National Coordinating Center (NCC) and Genetic Services Branch, MCHB Update***
Michael Watson, PhD, NCC, American College of Medical Genetics (ACMG)

2:20 pm – 2:40 pm ***"Leveraging a Regional Telehealth Network to Deliver to the Underserved"***
Rena Brewer, RN, MA, Southeastern Telehealth Resource Center

2:40 pm – 3:10 pm ***"Update on Medicaid & Reimbursement/policy"***
Mei Wa Kwong, JD, Center for Connected Health Policy

3:10 pm – 3:30 pm ***"Overview of Telehealth Resource Center-Texla"***
Billy U. Philips, Jr., PhD, MPH, Texla Telehealth Resource Center

3:30 pm – 4:00 pm **Break – SALON A FOYER**

4:00 pm – 4:50 pm ***"Expanding Telegenetic Services in the Southeast Region"***
Chair: Hans Andersson, MD, Tulane University
Participants:
Michael Adcock, MS University of Mississippi Medical Center
Suleima Salgado, MBA, Georgia Department of Public Health
Roberto T. Zori, M.D, University of Florida
Michael J. Lyons, MD, Greenwood Genetic Center
Meagan Farmer, MS, CGC, MBA, University of Alabama at Birmingham

4:50 pm – 5:20 pm "**Project ECHO"**
Sanjeev Arora, MD, MACP, FACG, UNM School of Medicine

5:20 pm – 5:50 pm **Keynote Address: *"Gene Sequencing and Newborn Screening"***
Robert L. Nussbaum, M.D., Invitae Corporation

5:50 pm – 6:10 pm ***"Natural History of MSUD in the NBS Connect Registry"***
Aileen Kenneson-Adams, PhD, MS, Emory University

6:10 pm – 6:30 pm ***"Implementation of NBS for Pompe and MPSI in Georgia"***
Patricia Hall, PhD, Emory Genetics Lab-Eurofins
Rossana Sanchez Russo, MD, Emory 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!

All Sessions and Workgroup Meetings are open to everyone!

Friday, July 20, 2018

7:00 am – 8:00 am **Industry Supported Symposium – Shire Medical Affairs – WINDSOR BALLROOM**
"Newborn Screening for Lysosomal Storage Disorders: Clinical Utility and the Road Ahead"
(Continental Breakfast included for attendees)

7:00 am – 8:30 am **Continental Breakfast – (for those not attending Symposium) - SALON B-C**

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

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: Pam Arn, MD - Nemours

8:45 am – 9:30 am **Invited Speaker: Marshall Summar, MD,** Chief, Division of Genetics and Metabolism, Children's National Health System, Washington, DC
"Rare Disease as an Emerging Field of Medicine"

9:30 am – 9:45 am ***"Establishment of a Developmental Neurogenetics Clinic: Characterization of a Cohort of 56 Consecutive Patients with Autism, Developmental Delay +/- Epilepsy"***
Hans Andersson, MD, Tulane University

9:45 am – 10:00 am ***"The Alabama Genomic Health Initiative: Design, Implementation, and Results from the First Year"***
Anna Hurst, MS, MD, University of Alabama at Birmingham

10:00 am – 10:15 am ***"Engaging Families in Care and in Improving Genetics Services"***
Beth Dworetzky, MS, Family Voices

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

10:45 am – 11:45 am **Platform Session 3 – SALON A**
Moderator: Neena Champaigne, MD – Greenwood Genetic Center

10:45 am – 11:00 am ***"Comparative Liver Pathology in Glycogen Storage Disease Type IIIA"***
Elizabeth Brooks, MS, DVM, Duke University Medical Center

11:00 am – 11:15 am ***"Serum BAP, CTX1, PTH, P1NP, and Vitamin D Association with DXA Outcomes and Dietary Patterns in Patients with Phenylketonuria (PKU)"***
Teresa Douglas, MS, PhD, Emory University Department of Human Genetics

11:15 am – 11:30 am ***"Development of an At-home Urine Test for Blood Phe-Level Monitoring for Phenylketonuria (PKU): Progress Update"***
Robert Latour, PhD, Clemson University

11:30 am – 11:45 am ***"GAII Clinical Symptoms and Metabolic Profiles in Patients without Confirmatory Molecular Genetic Findings"***
Kacie Riley, BA, MS, Duke University Health System

12:00 pm – 1:00 pm **Lunch – WINDSOR BALLROOM AND FOYER**

1:00 pm – 2:45 pm **Platform Session 4 – SALON A**
Moderator: Tim Wood, PhD – Greenwood Genetic Center

1:00 pm – 1:45 pm **Invited Speaker: Mark Dant,** Executive Director, Ryan Foundation
"Finding Tomorrow – The Journey to Treatment"

1:45 pm – 2:00 pm ***"Simply Test for MPS™ Enzyme-panel Program: A Testing Resource for Early and Accurate Diagnosis of Mucopolysaccharidosis Disorders"***
Tim Wood, PhD, Greenwood Genetic Center

2:00 pm – 2:15 pm ***"Expansion of Enzyme Testing in Dried Blood Spots (DBS) for the Diagnosis of 20 Lysosomal Storage Disorders"***
Laura Pollard, PhD, Greenwood Genetic Center

2:15 pm – 2:30 pm ***"Framework for Adding Newborn Screening Conditions in Florida"***
Scott Shone, PhD, RTI International

2:30 pm – 2:45 pm ***"The NC Nexus Project of Newborn Exome Sequencing"***
Cynthia Powell, MD, University of North Carolina at Chapel Hill

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

CONCURRENT INDUSTRY-SUPPORTED SYMPOSIUM

TIME	SWANNANOA ROOM	VICTORIA ROOM	WINDSOR BALLROOM
3:00 pm – 4:00 pm	***"New Treatment Option for Adults with PKU"*** – BioMarin Pharmaceutical Inc	***"Clinical Perspectives on Type 1 Gaucher Disease"*** - Ozlem Alpan, MD, Pediatrics, Lysosomal and Rare Disorders Research and Treatment Center, Fairfax, VA - Shire	***"Enhancing Patient Evaluations and NGS Results with Face2Gene: A Review and Mini Undiagnosed Case Forum"*** - FDNA
4:00 pm – 5:00pm			

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

SERN Concurrent Consumer Alliance Session
Moderator: Mary Lauren Salvatore, MPH, CHES, Emory University School of Medicine

8:00 am – 10:00 am	**Family Voices Training** – Beth Dworetzky	**ALEXANDER ROOM**
10:00 am – 10:15 am	**Break**	**ALEXANDER ROOM**
10:15 am – 11:00 am	**Successes in State PKU Organizations** – Alabama PKU Foundation; Georgia PKU Connect; Louisiana PKU Alliance; Tennessee PKU Foundation	**ALEXANDER ROOM**
11:00 am – 12:00 pm	**Taking a Coach-Approach to Parenting Children with Complex Healthcare Needs** – Elaine Taylor-Klaus, CPCC. PCC	**ALEXANDER ROOM**
12:00 pm – 1:00 pm	**Lunch and State Updates**	**ALEXANDER ROOM**
1:00 pm – 1:45 pm	**"Finding Tomorrow – The Journey to Treatment"** - Mark Dant, Ryan Foundation	**Salon A**
1:45 pm – 2:00 pm	**Break**	**TRAVEL TO ALEXANDER ROOM**
2:00 pm – 2:45 pm	**Panel for Consumer-Focused Groups** – Parent-to-Parent USA; Family Network on Disability; Family Voices; National Genetics Education & Family Support Center	**ALEXANDER ROOM**
2:45 pm – 3:15pm	**Emergency Preparedness: Accomplishments and Future Directions** – Frances Beasley, Shannon Holland	**ALEXANDER ROOM**
3:15 pm – 3:30 pm	**Closing Remarks**	**ALEXANDER ROOM**

All Sessions and Workgroup Meetings are open to everyone!

Saturday, July 21, 2018

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

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: Thomas Terrell, MD, Covenant Medical Group, Clinton, TN**
"The Role of Genetic Factors in Concussion Risk and Post-concussion Recovery in Athletes"

9:00 am – 9:15 am ***"Multi-system Effects of Trauma and Mitochondrial Dysfunction-associated PTSD in Mice"***
Graeme Preston, MS, Hayward Genetics Center, Tulane University

9:15 am – 9:30 am ***"Whole Genome versus Whole Exome Sequencing"***
David Bick, MD, HudsonAlpha Institute for Biology

9:30 am – 9:45 am ***"Clinical Grade Whole Genome Sequencing Data from Various Samples Types: Dried Blood Spots, Whole Blood, Saliva, and Tumor"***
Alexander Valencia, PhD, PerkinElmer Genomics

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

10:15 am – 11:00 am **Platform Session 6 – SALON A**
Moderator: Art Hagar, PhD – Georgia Department of Public Health

10:15 am – 10:30 am ***"First Year Experiences Performing Newborn Screening for Five Lysosomal Disorders in Tennessee"***
George Dizikes, PhD, Tennessee Department of Health

10:30 am – 10:45 am ***"Case Report: A Mosaic Ring Chromosome 13 with Segmental Duplication"***
Yuwen Li, MD, PhD, Hayward Genetics Center, Tulane University

10:45 am – 11:00 am ***"An AML Case Study: Elucidating the Significance of the Integrated Application of Karyotyping, FISH, and Microarray in Cancer Cytogenetics"***
David Nguyen, BA, Hayward Genetics Center, Tulane University

11:00 am – 11:15 am ***"Understanding Barriers to Genetic Testing for Sickle Cell Trait: The African-American Male Perspective"***
Shandrea Foster, BS, MS, University of South Carolina Genetic Counseling Program

11:15 am – 11:45 am **SERGG Business Meeting and Student Award Presentations –** Dan Sharer, PhD, Incoming President

11:45 am **Adjournment** – See you next year!

SAVE THE DATES

Future

SERN/SERGG ANNUAL MEETINGS

July 18-20, 2019

July 16-18, 2020

Renaissance Asheville Marriott Hotel

Asheville, North Carolina

PLATFORM PRESENTATIONS
(In order of presentation)

ESTABLISHMENT OF A DEVELOPMENTAL NEUROGENETICS CLINIC: CHARACTERIZATION OF A COHORT OF 56 CONSECUTIVE PATIENTS WITH AUTISM, DEVELOPMENTAL DELAY +/- EPILEPSY

Debbold E[1], Nelson S[2,3], Settles L[4], Morava E[1], Andersson HC[1]

[1]Hayward Genetics Center, [2] Dept of Pediatrics, [3] Dept of Neurology, [4]Center for Autism and Related Disorder, Tulane University Medical School, New Orleans LA

Interdisciplinary care and research models are effective to address the neurologic spectrum of autism/developmental delay/epilepsy. The evaluation of such patients can become extremely complex with varying perspectives of neuropsychologists, neurologists and geneticists. These three groups at Tulane collected data on a patient cohort seen by in an effort to characterize the most effective approaches to diagnosis and phenotyping. A total of 56 consecutive patients (43 male/13 female) were seen by all three groups. 50% of patients were first seen before age 6 years, while 50% were seen between ages 6-18 years. Patients were more likely to have height and weights in the top or bottom tenth percentiles for age. 24/56 Patients (43%) had a final dx of ASD. Patients with epilepsy (20/56) were more likely to have lower full-scale IQ than those with ASD only or ADHD/developmental delay only. Genetic testing for children in the cohort included variably testing for congenital diseases of glycosylation, aCGH, genomic epilepsy gene panels and metabolic screening. In total, 9 of 56 patients (16%) yielded pathogenic genetic testing results, while an additional 21 (38%) yielded abnormalities of unknown significance. The genetic test most likely to be abnormal was aCGH (7/46 abnl). 13/51 (25%) had abnormal brain MRI. 2/23 Patients had abnormal epilepsy gene panels (13/23 had VUS). Interaction between specialists of varying background allowed careful characterization of this extremely heterogeous population and may lead to refining of the diagnostic evaluation of ASD/developmental delay population with/without epilepsy.

THE ALABAMA GENOMIC HEALTH INITIATIVE: DESIGN, IMPLEMENTATION, AND RESULTS FROM THE FIRST YEAR

Hurst A[1], Cooper G[2] Moss I[1], Shaw B[1], Bowling K[2], Finnila C[2], May T[2], Nakano M[3], Sodeke S[4], Cimino JJ[5], Edberg J[6], Cannon A[1], East K[2], Cooper G[2], Fouad M[7], Curry W[8], Barsh G[2], Might M[9], Korf B[1]

[1]Department of Genetics, University of Alabama at Birmingham (UAB), Birmingham, AL; [2]HudsonAlpha Institute for Biotechnology, Huntsville, AL; [3]Department of Medical Education, UAB School of Medicine, Birmingham, AL; [4]National Center for Bioethics in Research and Healthcare, Tuskegee University, Tuskegee, AL; [5]Informatics Institute, UAB School of Medicine, Birmingham, AL; [6]Division of Clinical Immunology and Rheumatology, UAB School of Medicine, Birmingham, AL; [7]Office for Diversity and Inclusion, UAB School of Medicine, Birmingham, AL; [8]Primary Care and Rural Health, UAB School of Medicine, Birmingham, AL; [9]Personalized Medicine Institute, UAB, Birmingham, AL

The Alabama Genomic Health Initiative (AGHI) seeks to provide health benefits for participants while building a biobank for future research of genetic contributions to health and disease. Study design and funding utilized collaborative efforts from state legislature, academic, and nonprofit organizations for this five-year, $2 million/year project. We aim to enroll 10,000 general population individuals over five years for genotyping (with Illumina's Global Screening Array) and up to 150 individuals a year affected with undiagnosed genetic conditions for whole genome sequencing (WGS). In the first 11 months, AGHI enrolled 1,543 genotyping and 82 whole genome participants (32 families). Genotyping participants receive results of likely pathogenic or pathogenic findings from the ACMG Secondary Findings v2.0 list. The rate of reportable findings in completed testing is 1% (15/1434) with Sanger confirmation pending in 12 additional samples. WGS participants receive results related to their phenotype and secondary findings, if desired. Six of the first 29 WGS samples analyzed have a reportable finding (21%), including novel candidate genes. Participants complete health history questionnaires for genetic counselor review, and 680/1386 (49%) of participants reviewed to date were flagged to potentially benefit from additional communication. The study was carefully designed with an external oversight committee and internal working groups in bioethics, education, genomics, data/biobanking, and participant/provider engagement. Patient navigators facilitate recruitment and engagement, specifically reaching underserved populations. We engaged community leaders to build ethical trust and address stigma surrounding research participation. AGHI also provides education to primary care providers and genetic counseling to participants.

PLATFORM PRESENTATIONS
(In order of presentation)

ENGAGING FAMILIES IN CARE AND TO IMPROVE SERVICES

Frangenburg MI, Baker B, Dworetzky B
National Genetics Education and Family Support Center, Washington, DC

This session will focus on the work of the National Genetics Education and Family Support Center to partner with the RGN's to ensure that all individuals and families, especially those who are medically underserved, who have, or are at risk of having, a genetic condition, have the tools to be engaged in their own care, and have the opportunity to be meaningfully engaged in genetic service systems at a national, regional and local levels. The family center will share best practices for partnering and engaging with local, and state family organizations. We will provide examples of technical assistance available to RGN's and NCC, such as in-person trainings and webinars for families, individuals and self-advocates to support meaningful engagement in their own healthcare. Additionally, the family center will provide examples of training and webinars for RGN's and NCC geared towards increasing and assessing organizational capacity to engage diverse parent leaders and family organizations.

COMPARATIVE LIVER PATHOLOGY IN GLYCOGEN STORAGE DISEASE TYPE IIIA

Brooks ED[1,2], Yi H[1], Sun B[1], Lim J-A[1], Halaby C[1], Clinton LK[3], Mavis AM[1], Bangari D[4], Fyfe JC[5], Thurberg BL[4], Austin S[1], Kishnani P[1]
[1]Department of Pediatrics, Duke University Medical Center, Durham, NC; [2]Division of Laboratory Animal Resources, Duke University Medical Center, Durham, NC; [3]Department of Pathology, Duke University Medical Center, Durham, NC; [4]Department of Pathology, Genzyme, a Sanofi Company, Framingham, MA; [5]Laboratory of Comparative Medical Genetics, Michigan State University, East Lansing, MI

Glycogen Storage Disease type III (GSD III) is an autosomal, recessive disorder caused by a mutation creating deficiency of glycogen debrancher enzyme from mutations in the amylo-1,6-glucosidase (*AGL*) gene causing hepatic and muscular disorders in those affected. Progressive hepatic fibrosis is commonly seen in patients with GSD III and liver failure, hepatic adenomas and hepatocellular carcinoma has been reported in some cases. We had previously characterized the disease progression in curly-coated retrievers (CCR), a large breed canine model that proved difficult to maintain in a laboratory setting. We have created a more manageable canine model via crossbreeding the CCR dogs with smaller, mixed-breed dogs and generated a novel *AGL* KO mouse model via deletion of exons 6-10 from the *Agl* gene. Both the animal models developed similar liver pathologic features shown in human patients, such as elevated liver enzymes, increased hepatic glycogen content, and progressive hepatic fibrosis leading to cirrhosis. Hepatocellular adenoma was also described in an 18-month *AGL* KO mouse. These features correspond to findings of similar elevations in ALT and AST, as well as hepatic fibrosis in GSD III patients, in addition to a previously undescribed cohort of child and adult GSD III patients (n=11) with hepatic fibrosis. Robust animal models, such as these, can prove invaluable in the development of novel therapeutic agents to prevent hepatic fibrosis and other complications of GSD III.

PLATFORM PRESENTATIONS
(In order of presentation)

SERUM BAP, CTX1, PTH, P1NP, AND VITAMIN D ASSOCIATION WITH DXA OUTCOMES AND DIETARY PATTERNS IN PATIENTS WITH PHENYLKETONURIA (PKU)

Douglas TD[1], Goodman M[2], Coakley K[3], Singh RH[1]
[1]Metabolic Nutrition, Department of Human Genetics, Emory University, Atlanta GA; [2]MD/MPH program, Emory University School of Public Health, Atlanta GA; [3]Department of Individual Family and Community Education, University of New Mexico

BMD: Bone mineral density; BAP: Bone alkaline phosphatase; CTX: carboxy-terminal type 1 collagen crosslink; PTH: Parathyroid hormone; P1NP: Procollagen 1 Intact N-Terminal Propeptide; 25OHD: 25-hydroxy-vitamin D

OBJECTIVES: 1) Evaluate the role of bone turnover markers in DXA outcomes for patients with PKU 2) Determine impact of dietary factors on bone turnover in PKU. METHODS: Collected data from 96 males and females with PKU (age 4-54 yrs). An RD analyzed 3-day diet records using NDSR. Serum bone turnover markers were analyzed with immunoassay methods at the Maine Medical Center Research Institute: BAP and P1NP (bone formation), CTX (bone resorption), 25OHD and PTH (act in both capacities). Data was analyzed for pediatrics (≤ 17 yrs, n=61) and adults (18+ yrs, n=38) in SPSS 24 controlling for relevant cofactors (α=0.05). RESULTS: Several study participants had bone biomarker values outside reference range. DXA revealed ¼ of study group had BMD z-scores ≤ -1.0. Clinically low z-scores (≤ -2.0) occurred in three children and one adult. Pediatric z-scores inversely associated with CTX1, PTH, and P1NP; positively associated with 25OHD (p<0.04 for all); no correlation in adults. Other pediatric factors associated with bone turnover are plasma Phe, serum triglycerides, specific micronutrients, total sugar, kcal/kg, BMI, and % lean mass (p<0.05 for all). In adults other associated factors include linear age, sex, protein and folate intake, blood triglycerides, fasting glucose, and blood pressure (p<0.05). Biomarker associations held true when controlling for potential confounders. CONCLUSIONS: Serum bone turnover markers, affected by diverse factors, are important to BMD outcomes in patients with PKU. High prevalence of negative z-scores and serum bone markers outside reference ranges indicate pervasive risk of future bone disease. This analysis complements prior results published by Kathryn Coakley, PhD RDN (JIMD 2016).

DEVELOPMENT OF AN AT-HOME URINE TEST FOR BLOOD PHE-LEVEL MONITORING FOR PHENYLKETONURIA (PKU): PROGRESS UPDATE

Latour RA[1], Champaigne KD[2], Champaigne NL[3], Chumanov G[4], DeLuca JM[3,5], Douglas TD[6], Gurung SR[6], Korneva G[1], Lee EW[1], Singh RH[6], Smith JV[7], Wood T[3]
[1]Department of Bioengineering, Clemson University, Clemson, SC; [2]Circa Bioscience, LLC, Greenwood, SC; [3]Greenwood Genetic Center, Greenwood, SC; [4]Department of Chemistry, Clemson University, Clemson, SC; [5]School of Nursing, Clemson University, Clemson, SC; [6]Department of Human Genetics, Emory University School of Medicine, Decatur, GA; [7]SciTeck Diagnostics, Inc., P.O. Box 562, Arden, NC

At last year's SERGG meeting, we introduced our efforts on the development of a simple at-home urine test for monitoring blood phenylalanine (Phe) levels for individuals with PKU. Our method uses colorimetric coupons to measure the concentrations of phenylpyruvic acid (PPA) and creatinine (CRE), with urine PPA/CRE ratios shown to provide high correlation with blood plasma Phe levels over a range of 0 to 1,800 □M. Our original method used commercially available CRE test strips from SciTeck Diagnostics for CRE measurement. Despite the encouraging correlation, the data revealed a substantial degree of scatter. This scatter can largely be attributed to the inherent person-to-person variation in CRE production (related to muscle mass) as well as the use of the commercial test strips, which were designed by SciTeck for the purpose of detecting abnormally low CRE concentrations in urine (0 - 20 mg/dL), with the normal range of CRE in urine being 20 – 400 mg/dL. In attempt to reduce the scatter for our test method, we have been working with SciTeck's CRE reagents (provided through a collaboration with SciTeck Diagnostics) over the past year to develop our own CRE coupons that are designed for the full normal range of CRE. Testing is now ongoing using our redesigned CRE coupons combined with redesigned PPA coupons as well. With our new coupons, we anticipate a reduction in data scatter for improved correlation between urine PPA/CRE ratios and blood Phe levels, with application as an effective at-home Phe monitor for individuals with PKU.

PLATFORM PRESENTATIONS
(In order of presentation)

GAII-LIKE CLINICAL SYMPTOMS AND METABOLIC PROFILES IN PATIENTS WITHOUT CONFIRMATORY MOLECULAR GENETIC FINDINGS

Riley K, Pendyal S, Walley N, Young S, Koeberl D
Duke University Health System, Dept. of Pediatrics, Division of Medical Genetics, Durham, NC

Glutaric acidemia type II (GAII) is an inherited disorder that causes periodic metabolic acidosis, leading to episodes of weakness, lethargy, vomiting and hypoglycemia. There are two forms, with variable ages of onset, characterized by recognizable biochemical patterns. 75-87% of patients with biochemical manifestations of GAII have identified biallelic pathogenic variants in the *ETFA, ETFB* or *ETFDH* genes on targeted testing. Of the remaining cases, disease etiology remains elusive. Patient 1 was identified when he was hospitalized at age 12with severe nausea, vomiting and fatigue. Treatment with IV fluids, IV carnitine, and riboflavin was initiated after acylcarnitine profile (ACP) demonstrated marked elevations suggestive of GAII. Nausea and vomiting improved, however ACP abnormalities persisted. Targeted gene sequencing and deletion/duplication identified one pathogenic mutation in *ETFDH* (c.51dupT), however did not establish a molecular diagnosis of GAII, as a second mutation was not found. He continues to be treated with riboflavin supplementation and diet modification, recommended clinical treatment for GAII. His younger sister has a similar, milder presentation with identical genetic and biochemical abnormalities, while parents are unaffected. We subsequently identified, and present here, six additional patients (ages 10-63 years), with symptoms and ACP consistent with GAII, for whom targeted molecular testing is nondiagnostic. We hypothesize that this cohort represents a subset of patients with variable etiologies for GAII-like presentations, including but not limited to manifesting heterozygotes, synergistic heterozygosity and idiopathic GAII. Additional molecular and phenotypic studies on a larger cohort will be helpful for discovery of additional genetic loci for GAII.

SIMPLY TEST FOR MPS™ ENZYME-PANEL PROGRAM: A TESTING RESOURCE FOR EARLY AND ACCURATE DIAGNOSIS OF MUCOPOLYSACCHARIDOSIS DISORDERS

Clarke L[1], Cristian I[2], Pollard L[3], Wood T[3], Raggio C[4], Finch M[5], Jurecki E[5]
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The subtle, non-specific early signs/symptoms of mucopolysaccharidoses (MPS) present a diagnostic challenge. Clinicians may face challenges ordering an enzyme panel once a clinical suspicion of an MPS disorder has been established. Simply Test for MPS™ Enzyme-Panel Testing Program provides a no-cost option to streamline testing for United States patients presenting with signs/symptoms of MPS disorders. Testing kits include program details and instructions for specimen collection. Specimens are sent to Greenwood Genetics Laboratory for analysis, with results available within approximately 2 weeks. Since implementation in February 2017, >200 kits have been submitted for analysis from >100 institutions. Approximately 79% were requested by geneticists. The majority of referred patients were 0–4 years of age (43%) and 5–15 years of age (39%); 10% of tests represented patients >31 years of age. To date, there is an approximate 26% positive diagnostic yield rate for all MPS disorders (54/209) with the following distribution: MPS I (8), MPS II (7), MPS III (A:5, B:3, C:2), MPS IVA (18), MPS VI (10), and MPS VII (1). 72% of MPS IVA patients were noted to have vertebral body abnormalities that raised MPS suspicion and 44% were previously diagnosed with or possibly suspected of having spondyloepiphyseal dysplasia prior to testing. Simply Test for MPS™ has successfully facilitated the ability to confirm or rule out a diagnosis of MPS based on clinical signs/symptoms. Increased disease awareness and use of the program may lead to earlier diagnosis and management of MPS and ultimately improved patient outcomes.

PLATFORM PRESENTATIONS
(In order of presentation)

EXPANSION OF ENZYME TESTING IN DRIED BLOOD SPOTS (DBS) FOR THE DIAGNOSIS OF 20 LYSOSOMAL STORAGE DISORDERS

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Enzyme analysis is the gold-standard for the diagnosis of lysosomal storage disorders (LSDs). However, enzyme analysis in leukocytes requires that whole blood samples arrive to the testing laboratory within 24-48 hours of collection, and requires a relatively large volume of blood. This is problematic for the international shipment of specimens and for testing infants. The adaptation of these enzyme assays for use in dried blood spots (DBS) has ameliorated these issues for a large number of LSDs. Initially, enzyme analysis in DBS exclusively utilized 4-methylumbelliferone (4-MU) fluorogenic substrates. The limitation of this methodology is that each enzyme must be measured individually. The development of tandem mass spectrometry (MS/MS) substrates has allowed multiple enzyme reactions to be combined in a single reaction, increasing efficiency. We report validation results for a new 6-plex assay for the diagnosis of five Mucopolysaccharidosis (MPS) disorders (MPS II, IIIB, IVA, VI and VII) and neuronal ceroid lipofuscinosis type 2 in DBS using UPLC-MS/MS. Normal reference ranges were developed using a minimum of 236 DBS samples from unaffected controls or patients with an alternate diagnosis, and clinical sensitivity was established using 99 samples from patients affected with one of the six disorders. Intra-day and inter-day precision was acceptable (< 20% CV). Our laboratory can now analyze 18 enzymes in DBS (12 utilizing MS/MS substrates and 6 utilizing 4-MU substrates) to diagnose 20 different LSDs. Furthermore, we can now offer a 7 enzyme MPS panel in DBS, which will significantly enhance the worldwide diagnosis of MPS patients.

FRAMEWORK FOR ADDING NEWBORN SCREENING CONDITIONS IN FLORIDA

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The Secretary of Health and Human Services's Advisory Committee on Heritable Disorders in Newborns and Children (ACHDNC) has developed a thorough and rigorous evidence review process to evaluate disorders that are nominated for addition to the Recommended Uniform Screening Panel. The Florida Department of Health contracted with RTI International to develop a similar framework for the addition of conditions to the state's mandated screening panel. The project resulted in two reports that proposed specific review criteria and a framework under which to consider these criteria when evaluating new conditions. The RTI project team evaluated relevant peer-reviewed publications for newborn screening disorder review processes utilized at the national level, as well as publications from states and international newborn screening programs. In addition, an environmental scan was performed to examine scientific presentations at national and international conferences and to evaluate newborn screening program websites for disorder review information. An initial report summarized proposed review criteria in four categories (Screening Test, Benefits of Early Detection, Public Health System Readiness, and Cost). A subsequent report described the applicability of the ACHDNC framework for each review criterion and detailed additional state-specific steps that would inform the disorder review process. Florida's Genetics and Newborn Screening Advisory Council were in favor of using the proposed criteria and framework at their February 2018 meeting. Details from the framework reports will be presented.

PLATFORM PRESENTATIONS
(In order of presentation)

THE NC NEXUS PROJECT OF NEWBORN EXOME SEQUENCING

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The North Carolina Newborn Exome Sequencing for Universal Screening (NC NEXUS) research project is examining the utility of and parental decision-making for genomic sequencing in newborns and children. Two cohorts of patients are included: one from birth to age 5 years with known conditions diagnosed through standard newborn screening and a second of healthy newborns whose parents are recruited during the prenatal period. Both cohorts receive results for pathogenic variants in genes associated with conditions determined to have childhood onset and medical actionability (called the Next-Generation Sequencing-Newborn Screening or NGS-NBS category). There are currently 466 gene-disease pairs in this category. Results: 541 individuals were approached to participate; 356 (66%) agreed to join the study. Of those who joined the study, 96 (27%) consented to having their child's genome sequenced. Sequencing has been completed in 63 cases. Of 15 cases sequenced in the metabolic cohort, the known metabolic disorder was confirmed in 13. In one female patient with known PKU, a likely pathogenic variant was also identified in the ornithine transcarbamylase (*OTC)* gene. Of 22 patients sequenced thus far in the hearing loss cohort, a genetic etiology was identified in 23%, including Pendred syndrome, Usher syndrome type 2A (2 cases), non-syndromic hearing loss due to *GJB2* and *TECTA*. Two cases in this cohort had a positive, unrelated finding in the NGS-NBS category. Of the 25 healthy newborns sequenced, none have had findings in the NGS-NBS category. Of children sequenced thus far, 3% have had a significant finding in the NGS-NBS category.

MULTI-SYSTEM EFFECTS OF TRAUMA AND MITOCHONDRIAL DYSFUNCTION-ASSOCIATED PTSD IN MICE

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Mitochondrial dysfunction has been increasingly implicated in several psychopathologies, and mitochondria play a central role in several physiologic processes associated with the pathophysiology of PTSD. We recently showed that mitochondrial dysfunction in the brain significantly correlated with susceptibility to Post-Traumatic Stress Disorder in mice. Here, we assess the physiologic effects of trauma and PTSD-susceptibility across several systems in these animals.
PTSD was induced in a cohort of WT FVB mice, and PTSD-vulnerable and resilient animals were identified. The activities of the mitochondrial electron transport chain (ETC) complexes I, II, III and IV isolated from brain and muscle of PTSD vulnerable and resilient animals, were measure. Blood plasma was collected and ELISA for Corticosterone and FGF-21 was performed. Thymi were fixed in 0.4% PFA, sectioned, and stained with hematoxylin and eosin, and physiology was assessed.
Muscle mitochondrial ETC activity did not vary significantly between PTSD-vulnerable and resilient animals, and was not correlated with PTSD vulnerability. Traumatized and PTSD-vulnerable mice showed a significant decrease in the liver stress hormone FGF-21, but not the adrenal stress hormone corticosterone. PTSD vulnerable animals also thymic structural changes consistent with thymic involution and chronic stress.
Our data shows that the mitochondrial dysfunction that correlates with PTSD vulnerability in these animals is tissue and brain specific. We also found evidence of trauma and PTSD-induced dysfunction in another highly-metabolic tissue -- the liver – as well as evidence of chronic stress associated with PTSD-vulnerability. These data may help to illuminate new avenues for diagnosis, treatment and prevention of PTSD.

PLATFORM PRESENTATIONS
(In order of presentation)

WHOLE GENOME VERSUS WHOLE EXOME SEQUENCING

Bick DP
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Genomic testing has revolutionized the clinician and laboratory's ability to care for patients with rare disorders by increasing diagnostic yield and shortening time spent in a diagnostic odyssey. Exome and genome sequencing technologies are often discussed and considered together. However, this can minimize critical differences between the technologies that should be understood when determining which is most useful for a particular case. This session will compare technical aspects of exome and genome sequencing including analytical and clinical validity. Genome sequencing has been shown to outperform exome sequencing in identifying a full spectrum of genetic changes, including copy number changes and trinucleotide repeat expansions. Genome sequencing also provides the most complete coverage of exons –making it the best way to interrogate the exome. Implementing genome sequencing increases diagnostic yield while also providing a superior dataset for future reanalysis. In addition to technical features, cost and reimbursement must be considered with both exome and genome sequencing. As the price of genomic sequencing decreases and the availability of genomic research expands, more patients have access to whole exome and genome sequencing tests than ever before. The particular tests available to a patient in a given setting is highly influenced by cost and payer reimbursement or other funding. With a growing number of national and state funded initiatives acknowledging the importance of providing genomic information to patients, cost and access paradigms are shifting. This session will highlight these paradigms and the future directions for genomic testing in the clinical and research setting.

CLINICAL GRADE WHOLE GENOME SEQUENCING DATA FROM VARIOUS SAMPLES TYPES: DRIED BLOOD SPOTS, WHOLE BLOOD, SALIVA AND TUMOR

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Whole genome sequencing (WGS) is becoming increasingly attractive as an alternative to other molecular methods, due to its broader coverage and decreasing cost as well as higher sensitivity for the detection of variants. Providing sample type options, dried blood spots (DBS), saliva, whole blood (WB, EDTA) and tumor to patients may increase the participation in genetic testing. The objective of this study was to assess whether a variety of sample types provide clinical grade whole genome data by comparing the global metrics and performance parameters. In the experimental design, we sequenced the whole genome without DNA amplification from three healthy adult individuals that provided DBS, saliva, and WB, along with control DNA (NA12878) and a tumor sample, on the NovaSeq 6000 using S2 flowcells. The coverage of the genome, total number of SNPs, total number of indels, number of heterozygous and homozygous calls was similar for the four sample types. Based on the WGS of NA12878 compared to genome in a bottle, the accuracy was calculated to be 98.90% by including all SNPs. The precision, sensitivity and specificity values were comparable for the three sample types. This evidence supports that properly stored DBS, saliva, blood and tumor were all satisfactory sources of DNA for WGS studies using Illumina NovaSeq technology. The viability of DBS as a source of quality DNA opens up the possibility that next-generation sequencing can play a role in newborn sequencing.

PLATFORM PRESENTATIONS
(In order of presentation)

FIRST YEAR EXPERIENCES PERFORMING NEWBORN SCREENING FOR FIVE LYSOSOMAL DISORDERS IN TENNESSEE

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The Tennessee Department of Health Newborn Screening Laboratory began state-wide screening for five lysosomal disorders on July 1, 2017, two years after recommendation by their Genetics Advisory Committee and endorsement by the Commissioner. Testing for Pompe, Mucopolysaccharidosis Type I, Gaucher, Fabry, and Krabbe Diseases was performed by tandem mass spectrometry using reagents developed by Perkin-Elmer, Inc. After method validation, approximately 70,000 specimens were tested during the first nine months of screening. Overall 122 patients were referred to short-term follow-up, and 16 of these were subsequently confirmed positive. During this time, initially conservative cutoff values were refined to reduce false-positive call outs, and presumptively positive specimens were sent out for second-tier biochemical testing and DNA sequence analysis in order to further improve specificity and disease prognosis. In addition, the Mayo Clinic method of CLIR analysis was applied to these data in order to also improve specificity and positive predictive values. Evaluation of these higher tier tests and comprehensive methods of data analysis were evaluated with regard to their effects on overall testing and follow-up costs, turnaround times, and patient outcomes. Biochemical second-tier tests, available for all the disorder except Fabry, promise to be cost-effective, rapid methods to rule out false-positive screening results – thereby reducing the number of call-outs and the time and expense of unnecessary follow-ups. Conserved resources subsequently applied to DNA analysis can help to define a diagnosis during the newborn period, providing the opportunity for more effective intervention and eliminating the need for a costly and lengthy diagnostic odyssey.

CASE REPORT: A MOSAIC RING CHROMOSOME 13 WITH SEGMENTAL DUPLICATION

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Our patient was prenatally suspected to have trisomy 13 via non-invasive prenatal testing (NIPT). This was determined to be a mosaic ring chromosome 13 by amniocentesis at a different facility. Labor was induced at 39 weeks due to maternal hypertension, and he presented with a weak cry, central cyanosis and respiratory congestion at birth. At four-months, he presented at clinic with developmental delay, hypotonia, broad nasal bridge and mild micrognathia. Otherwise, he was normal without typical features of trisomy 13. Chromosomal karyotyping of peripheral blood identified a mosaic ring chromosome 13 including a single ring 13 p13q34 (44 cells), monosomy 13 (5 cells), and a double/dicentric ring 13 (3 cells), in 52 cells examined. A segmental duplication was suspected in ring 13 structure therefore a CGH+SNP array was performed to confirm the duplication and further characterize the break points. The array revealed a 23.55 Mbp duplication of the 13q31.2q34 segment, as well as a 590 Kbp terminal deletion from 13q34. According to the genomic coordinates of the duplicated segment acquired by CGH, two BAC FISH probes (RP11-625H9 - GREEN and RP11-106A13 - RED) were selected from the duplicated region to define the orientation of the duplication, which confirmed an inverted duplication. By using comprehensive genetic and genomic analyses, we identified a mosaic ring chromosome 13 with 13q31.2q34 inverted duplication in a patient with developmental delay. Our literature search suggests that this is only the third case of ring 13 with segmental duplication reported.

PLATFORM PRESENTATIONS
(In order of presentation)

AN AML CASE STUDY: ELUCIDATING THE SIGNIFICANCE OF THE INTEGRATED APPLICATION OF KARYOTYPING, FISH, AND MICROARRAY IN CANCER CYTOGENETICS

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Acute myeloid leukemia (AML), a cancer of blood and bone marrow, is marked by a high degree of heterogeneity in gene mutations and chromosomal abnormalities. Identification of these aberrations plays an integral role in terms of diagnostics, prognostics, and treatment. Traditionally, these aberrations have been detected with cytogenetics and fluorescence in situ hybridization (FISH), which have limitations in resolution and sensitivity. Newer technologies, such as array comparative genomic hybridization (aCGH) + Single Nucleotide Polymorphism (SNP) microarray have improved the detection of genomic variations in myeloid leukemias. Here we report the karyotype, FISH, and microarray findings of a patient with AML with AML, in order to elucidate the significance of the integrated analytic strategy. Bone marrow aspirate from a 39-year old female was received for cytogenetic and FISH analysis. G-banding revealed several structural and numerical abnormalities involving chromosomes 1, 5, 17, 20, and 21, as well as a marker chromosome. FISH further identified deletions in CKS1B, EGR1, and TP53, as well as RUNX1 amplification. Microarray studies were then performed to further characterize the abnormalities. These results confirmed the abnormalities seen in chromosomes 5, 17, and 20, and helped to characterize the aberrations involving chromosomes 1 and 20, 5 and 17, the marker chromosome, as well as the 21q amplification. In this case report, the extensive genome-wide analyses enabled us to depict the chromosomal aberrations more precisely and suggest a reciprocal complementary strategy of integrating G-banding, FISH, and microarray analyses in cancer cytogenetics application.

UNDERSTANDING BARRIERS TO GENETIC TESTING FOR SICKLE CELL TRAIT: THE AFRICAN-AMERICAN MALE PERSPECTIVE

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Research has shown a reluctance in African-American males to pursue testing for sickle cell trait. Few studies have tried to discern what barriers are contributing to this issue within the African-American male community. Research suggests a lack of knowledge may be the biggest contributing factor. This study hypothesized there would be a significant difference in knowledge of sickle cell trait based on educational level, age, and health beliefs. African-America male participants (N=116), ages 18 and over, completed a questionnaire assessing knowledge, risk perception, health beliefs, barriers, and motivating factors within the context of sickle cell trait. One-way and two-way analysis of variance identified age as an influential factor. Results showed a significant interaction between age and knowledge of sickle cell trait and sickle cell disease ($p = .009$). Factors including perceived discrimination, perceived risk of sickle cell trait based on parent report, and sentiments on playing sports with sickle cell trait were all influenced by age (all $p < 0.05$). Health beliefs such as having tattoos or piercings and getting annual check-ups with a primary care physician were also influenced by age (both $p < 0.02$). The most significant barrier identified was a lack of information about testing options from primary care physicians, while the largest motivating factor for testing was for personal health reasons. Findings from this study could aid genetic counselors with strategies to increase sickle cell trait testing in African-American men. Thereby, increasing awareness of sickle cell trait in the community for informative health and reproductive outlook.

POSTER PRESENTATIONS
(Alphabetical by presenting author)

APPROACHES TO TELEMEDICINE CLINIC FOR METABOLIC PATIENTS

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Successful treatment of patients with metabolic disease is based around the issues of compliance with treatment and compliance with monitoring. The University of Florida has instituted telemedicine follow-up for patients with inborn errors of metabolism via quarterly telemedicine clinic appointments and monthly telemedicine home visits. Clinic visits are held quarterly with dietitians providing nutritional counseling, while these patients receive on site genetics consultation at satellite CMS clinics. Home visits are provided via telemedicine in a patient's home, by specialists at the University of Florida Division of Pediatric Genetics and Metabolism. This program is currently being utilized for new and return patients. The application Vidyo is being used for web conferencing. This allows our division to see patients more efficiently and effectively as many of our patients live over 3 hours from our main clinic site. The purpose of this program is to bridge the gap and provide equal access to care to all patients, despite growing differences in demographics. Appropriate consents for telemedicine evaluation are obtained prior to the evaluation. Medical and developmental history is obtained from our center, newborn screening documents, and pediatrician notes. In these clinic visits we provide nutritional and medical evaluation along with counseling from our distant site. If necessary follow up appointments can be made for the following week. This poster will detail the methodology of metabolic telemedicine clinics at the University of Florida, a metabolic case report, and barriers to providing care through telemedicine.

WOMEN, INFANTS, AND CHILDREN OF GEORGIA AND COVERAGE OF MEDICAL FORMULA IN INBRON ERRORS OF METABOLISM: POLICIES, PROCEDURES, AND OVERAGE CONVERAGE BY MEDICAL NUTRITION THERAPY FOR PREVENTION PROGRAM

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BACKGROUND: Medical foods (MF) are essential for meeting nutritional needs in patients with inborn errors of metabolism (IEM). High cost and inadequate insurance reimbursement of MF places significant financial burden on individuals and families affected by IEM. In Georgia, the start-up MNT4P program assists families with MF overage coverage challenges while documenting the scope of overage coverage gaps for MF. OBJECTIVE: We investigated the current Georgia WIC policies as related to the approval and coverage of MF for families with children age ≤ 5 years diagnosed with an IEM. We also evaluated costs of covering the expense gap for MF by the MNT4P program given current limitations in WIC coverage. METHODS: A focused interview was conducted with officials at Georgia WIC to better understand the Georgia WIC criteria for approving and adding metabolic formulas to the exemption and how formula package maximums are calculated. Total WIC cost and overage coverage cost was acquired from January 2017 WIC/MNT4P cohort data, age ≤ 5 years (N=28). RESULTS: It was found that 36% of patients required overage coverage with 90% of those requiring overage coverage diagnosed with an IEM other than PKU. On average, having an IEM other than PKU increases the risk of having overage costs by 5.82 times ($p=0.04$). CONCLUSIONS: More than 1/3 of IEM patients have significant expense coverage gaps for essential MF via WIC or other means; the majority affected being those with non-PKU disorders. Evaluation of overage coverage across a longer time in the WIC/MNT4P cohort is ongoing.

POSTER PRESENTATIONS
(Alphabetical by presenting author)

THE SUPER RESPONDERS: KUVAN PROVIDES SIGNIFICANT DIETARY LIBERALIZATION IN 18 OF 72 PKU PATIENTS

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*Previous studies have shown Kuvan (Sapropterin) therapy reduces plasma phenylalanine (Phe) in some patients with Phenylketonuria (PKU). A subgroup of Kuvan responders may significantly liberalize dietary Phe intake while maintaining blood Phe control. Our clinic trialed 72 patients on Kuvan and 41 of these patients were considered responders (57%). Eigh*teen increased daily Phe tolerance two fold or more while taking Kuvan. *A retrospective chart review was conducted on 72 PKU patients trialed on Kuvan at dosages of 20 mg/kg daily for four weeks. When blood Phe response occurred, a systematic dietary Phe challenge measured increased tolerance. Patients with a drop in blood Phe levels of 30% or greater within the first trial week, and at least 2 fold increase in dietary Phe intake were labeled "super-responders" and included in our analysis. A paired one-tail student t-test measured statistical significance (P ≤0.05). Eighteen patients with PKU were included in our study. All were Caucasian and aged 5-39 years. Mean Phe intake increased by 322% while maintaining mean plasma Phe levels within treatment range (120-360 umol/L). This treatment outcome has continued over time. 25% of our PKU patients trialed on Kuvan had a rapid and significant decrease in blood Phe and an increase in dietary Phe tolerance of at least 2 fold, classifying them as "super-responders". All reported a significant improvement in quality of life.*

THE IMPACT OF GENETIC DISEASE IN GENERAL INPATIENT PEDIATRIC MEDICINE

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Individually genetic disorders are rare, but collectively genetic disorders are common. To determine the impact of genetic diseases on the General Inpatient Pediatric Medicine service at Children's of Alabama in Birmingham, AL, a retrospective chart review was conducted. This retrospective review determined that for the month of January 2018, 15% of all admissions to a General Inpatient Pediatric Medicine team were due completely to genetic diseases and birth defects. Admission diagnoses ranged from Niemann-Pick disease type C to X-linked chronic granulomatous disease to Trisomy 21. We compared our data to prior studies, and the results were consistent with prior studies. In particular, a 1997 study by Yoon et al that found that 12% of pediatric hospitalizations were related specifically to genetic diseases and birth defects. Our results were also relatively comparable to a landmark study published in 1978 by Hall et al that revealed that greater than 50% of pediatric hospital admissions were due to a genetically determined condition. Specifically, 4.5% were due to completely genetic conditions, 22% were due to multifactorial conditions and 27% were due to developmental or familial disorders. The data from Children's of Alabama continues to support the tremendous impact that genetic diseases have on general inpatient pediatric medicine. We plan to repeat the review for different inpatient teams and months to gather additional data and assess for seasonal variation.

POSTER PRESENTATIONS
(Alphabetical by presenting author)

MARKERS OF PROTEIN TURNOVER ACROSS ONE YEAR IN PATIENTS WITH PHENYLKETONURIA (PKU) ON STANDARD AND RELAXED DIET

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OBJECTIVES: 1) Compare protein turnover markers in healthy controls to those with PKU to assess distinctions in protein metabolism 2) Determine protein turnover impact of increased dietary Phe tolerance and intact protein intake among PKU sapropterin responders. METHODS: Baseline data collected from male and female patients with PKU age 4-50 yrs (n=58) and healthy controls age 7-37 yrs (n=13). An RD analyzed 3-day diet records using NDSR. The PKU cohort returned for follow up visits across one year. At each visit, serum and 12-hour overnight urine samples were analyzed for protein markers including creatine and creatinine. Results was compared across time and groups with parametric regression methods in SPSS 24 (α=0.05). RESULTS: Pediatric PKU patients had urine creatinine excretion almost half of controls (mean difference: 85.7mg/dL ± 20.5 SD, p=0.003). Significance remained when incorporating linear age and urine volume. All other lab markers were equivalent to controls. Creatinine and creatine did not change across one year regardless of diet or sapropterin response. Urinary creatinine and serum creatine correlated in adults (p=0.001) but not children with PKU. In PKU children, urine creatinine associated marginally with g/kg/day total dietary protein and kcal/kg/day (p=0.07). Kidney function markers (eGFR, BUN) were within reference range throughout study for all participants. CONCLUSIONS: In PKU youth, lower urine creatinine compared to controls could indicate alterations in protein metabolism. Diet liberalization across one year in sapropterin responders did not alter protein turnover markers, however associations with protein and energy intake demonstrate an extent of dietary influence.

ENGAGING FAMILIES IN CARE AND TO IMPROVE SERVICES

Frangenburg MI, Baker B, Dworetzky B
National Genetics Education and Family Support Center, Washington, DC

This session will focus on the work of the National Genetics Education and Family Support Center to partner with the RGN's to ensure that all individuals and families, especially those who are medically underserved, who have, or are at risk of having, a genetic condition, have the tools to be engaged in their own care, and have the opportunity to be meaningfully engaged in genetic service systems at a national, regional and local levels. The family center will share best practices for partnering and engaging with local, and state family organizations. We will provide examples of technical assistance available to RGN's and NCC, such as in-person trainings and webinars for families, individuals and self-advocates to support meaningful engagement in their own healthcare. Additionally, the family center will provide examples of training and webinars for RGN's and NCC geared towards increasing and assessing organizational capacity to engage diverse parent leaders and family organizations.

POSTER PRESENTATIONS
(Alphabetical by presenting author)

LONG TERM SAFETY AND EFFICACY OF GLYCEROL PHENYLBUTYRATE FOR THE MANAGEMENT OF UREA CYCLE DISORDER PATIENTS

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Introduction. Glycerol phenylbutyrate's (GPB) safety and efficacy has previously been determined in 3 short-term switch-over studies and their long-term 12 month follow-up analyses. Herein, we report additional longer term data on this population out to 54 months. Methods. This was an open-label, long-term study of 88 UCD patients who received GPB at the same maintenance dose as the previous enrolled study. Normalized (n) plasma ammonia levels (including those above the upper limit of normal (nULN)) and number of hyperammonemic crises (HACs) were assessed at baseline and every 6 months. AEs, vital signs, and clinical laboratory measurements were monitored. Results. 82 patients (43 pediatric and 39 adult) completed the study. Three patients were lost-to-follow up, 1 withdrew, 1 relocated, and 1 underwent a liver transplantation. The median duration of treatment was ≈ 1.85 years in this follow-up study with a duration of treatment inclusive of the previous enrolled GPB studies of ≈ 2.84 years. Mean ammonia levels were below the nULN (<35 µmol/L) until month 48, where there was increasing ammonia fluctuations, likely due to the small number of patients at months 54 (n=5) to 66 (n=2). Glutamine levels significantly decreased over time. Annualized HAC rates were lower with GPB as compared with pre-enrollment in the overall group and in both pediatric and adult populations. No new safety signals were identified in this long-term follow-up study. Conclusion. UCD patients treated with GPB and followed over a median of 2.8 years maintained ammonia control with no new adverse events identified.

THE PROVISION OF TELEMEDICINE GENETIC SERVICES TO TWO DIFFERENT SITES IN FLORIDA

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The birth of a baby with multiple congenital anomalies (MCAs) or positive prenatal genetic test can provide many management and diagnostic challenges for a neonatal intensive care unit (NICU). Access to consultation with a geneticist and genetic counselor provides many benefits for both the medical team and the family. However, in underserved regions, including some with major hospitals, genetics services are not available. The use of telemedicine has allowed us to provide real time genetics consultations to two different locations: Sacred Heart Hospital in Pensacola, and Wolfson Children's Hospital in Jacksonville, FL. We additionally provide consultation with the Child Neurology Center in Gulf Breeze, FL. We utilize HIPAA compliant telemedicine software called Vidyo to perform full genetics consultations via iPad. These consultations consist of obtaining a prenatal history and detailed pedigree, a full guided physical exam, discussion of the differential diagnosis, management recommendations, guidance on the most appropriate genetic test, and help with ordering the testing. We then meet with the family to perform genetic counseling regarding the diagnosis or differential diagnosis, the potential recurrence risk(s), and to obtain informed consent for the genetic testing. This program results in significant cost savings by identification of the most appropriate and targeted genetic testing and provides the family with a better understanding of the genetic concerns or diagnosis, and recommended genetic testing and anticipatory guidance. We will describe our experience with telemedicine consults, and present data on the improved outcomes.

POSTER PRESENTATIONS
(Alphabetical by presenting author)

IDENTIFYING CAUSATION THROUGH THE UNDIAGNOSED DISEASES NETWORK (UDN): A LIMB-GIRDLE MUSCULAR DYSTROPHY CASE STUDY

Koziura ME[1], Fairbrother L[1], Cogan J[1], Rives L[1], Bican A[1], Pfotenhauer J[1], Mobley B[3], Bhatia A[4], Markham L[1], Brault J[1], Phillips JA[1], Newman J[2], Hamid R[1].
Departments of Pediatrics[1], Medicine[2], Pathology[3], and Radiology[4], Vanderbilt University Medical Center, Nashville, TN

Background: According to the National Organization for Rare Diseases (NORD) 30 million Americans are living with rare diseases. The majority of them are currently undiagnosed. This case study will highlight how utilization of clinically available testing methods along with research genetic testing can identify the etiology for an individual's constellation of symptoms. Methods: First, Whole Genome Sequencing (WGS) was performed through the Undiagnosed Diseases Network (UDN). WGS revealed two mutations in TOR1AIP1 (c.554-4G>A & 1427C>T) classified as likely pathogenic. Second, histological studies and radiological imaging was performed during the proband's in-person evaluation to aid in determining the significance of these findings from other research candidates of interest. Third, cDNA was also performed. Findings: Sanger sequencing for both mutations in TOR1AIP1 was confirmed. Radiological imaging showed findings consistent with TOR1AIP1 as did the histological studies. cDNA confirmed pathogenicity by disrupting the splice site. Conclusion: The use of both clinical diagnostic testing and research testing allowed for the UDN to provide a diagnosis for a 17 year old female that had been evading her providers for over 9 years. As the field of Genetics and Genomics continues to advance in not only its knowledge of rare diseases, unique presentations of known diseases, and technology in confirming pathogenic etiologies for our proband's clinical phenotype it will continue to be an adjunctive measure to our clinical diagnostic tools, thereby, allowing providers to make the necessary associations to diagnose our patients and contribute further knowledge to the science of medicine.

KNOWLEDGE AND PERCEPTIONS OF ASSISTED REPRODUCTIVE TECHNOLOGIES AND OTHER FAMILY BUILDING OPTIONS IN ADULTS WITH CYSTIC FIBROSIS

Kushary S[1], Ali N[1], Spencer J[2], Dokson J[3], Hunt WR[4]
[1]Department of Human Genetics, Emory University, Atlanta, GA; [2]Department of Gynecology and Obstetrics, [3]Reproductive Biology Associates, Atlanta, GA; [4]Department of Medicine, Emory University, Atlanta, GA

Cystic fibrosis (CF) is a congenital lung disease affecting over 30,000 people in the United States. Advances over the last 40 years have increased life expectancy of patients, leading to an increased desire for adult patients to become parents. Patients should be fully informed about the genetics of CF and family building options available to them such as assisted reproductive technologies before pursuing parenthood. This study aims to learn about the current knowledge level and the personal perceptions about genetics and assisted reproductive technologies that exist in this patient population. In addition, it aims to see how a genetic counseling session may impact knowledge and perception. The study population includes individuals over the age of 18 who are currently seen at the Emory Adult Cystic Fibrosis Clinic. A combination of pre-session and post-session surveys will be used to assess topics related to genetics and assisted reproductive technologies. Each patient will be asked to take the survey before the genetic counseling session, immediately after the session, and 4 to 8 weeks after the session. Difference in scores between the questionnaires will be analyzed to see if knowledge and perception answers have changed after the session. We will also assess how demographic factors affect these scores. The results of this study will inform us about the knowledge gaps and current perceptions present in this patient population. Furthermore, it will evaluate how genetic counseling may affect both of those things, impacting how we may care for future generations of patients with cystic fibrosis.

POSTER PRESENTATIONS
(Alphabetical by presenting author)

NORTH CAROLINA X-LINKED ADRENOLEUKODYSTROPHY (X-ALD) PILOT STUDY

Lee S[1], Taylor J[1], Shone S[1], Beckloff S[2], Patel H[2], Zimmerman S[2], Young S[3], Millington D[3], Rehder C[4], Clinard K[4], Powell C[4], Bailey D[1].
[1]RTI International, Research Triangle Park, NC; [2]North Carolina State Laboratory of Public Health, Raleigh, NC; [3]Duke University Biochemical Genetics Laboratory, Durham, NC; [4]University of North Carolina, Chapel Hill, Chapel Hill, NC

X-ALD is an X-linked, recessive genetic disorder caused by mutations in the *ABCD1* gene. Mutations in *ABCD1* result in peroxisomal dysfunction and accumulation of very long chain fatty acids (VLCFAs) in all tissues of the body. However, X-ALD primarily manifests in the adrenal cortex and white matter of the nervous system. X-ALD is the most common peroxisomal disorder, with an estimated incidence of 1 in 14,700 live births in the U.S. In 2016, X-ALD was added to the Recommended Uniform Screening Panel (RUSP); however, only a few states have implemented full population screening. The National Institute for Child Health and Human Development awarded a contract to RTI International (RTI), the University of North Carolina Chapel Hill, Duke University, and the North Carolina State Laboratory of Public Health to conduct a newborn screening pilot study for X-ALD. RTI and NCSLPH implemented a stand-alone test using negative ion mode HPLC-MS/MS to determine lysophosphatidylcholine levels in dried blood spots. Specimens with significantly elevated levels of C26:0-LPC or moderately elevated levels of C26:0-LPC and elevated levels of C24:0-LPC were considered screen positive and referred for follow-up. Specimens with moderately elevated levels of C26:0-LPC and low levels of C24:0-LPC were considered borderline and an additional specimen was requested for reevaluation. Infants with screen positive results received confirmatory testing including *ABCD1* gene sequencing and VLCFA analysis. The X-ALD pilot study began on March 5th, 2018 and included 50,000 specimens. Screening results and diagnostic confirmation data for screen-positive specimens will be presented.

MANAGEMENT OF PATIENTS WITH MILD CITRULLINEMIA DIAGNOSED VIA NEWBORN SCREEN: A CASE SERIES

Meyer-Hacker SB, Ragin-Dames L, Tekin M
Division of Clinical and Translational Genetics, University of Miami Miller School of Medicine, Miami, FL

Citrullinemia type 1 is a urea cycle defect caused by a deficiency of Arginosuccinate Synthatase (ASS) which presents on a clinical spectrum. While there are published guidelines for the management of symptomatic forms of the condition, the published literature provides little guidance for the management of patients with mild presentations. We present a case series of three patients diagnosed with Mild Citrullinemia. Patients are currently aged 2 months-3 years. Patients were ascertained via Newborn Screen. Confirmatory testing was completed within the first 3 weeks of life in all subjects. Citrulline was between 123-198 uMol/L and Ammonia between 43-78 uMol/L. All subjects were found to have two mutations in the ASS1 gene, with at least one known pathogenic variant. At the initial visit, all subjects were educated about the condition and provided with a sick day protocol. All patients were breastfed, with one subject receiving supplementation of standard infant formula. Patients continued with normal protein diets, with one patient receiving a small amount of an essential amino acid formula. There have been no hyperammonemic episodes reported in any patients, even through illness. Patients all have normal growth and development. They continue to be frequently monitored as risk of hyperammonemia is not known in this population. Our experience highlights the need for a better understanding of the mild forms of Citrullinemia to more safely and appropriately monitor and manage affected patients.

POSTER PRESENTATIONS
(Alphabetical by presenting author)

NEWBORN SCREENING: A COMPREHENSIVE MODEL FOR MEDICAL NUTRITION THERAPY

Salvatore ML, Gurung S, Singh RH
Emory University School of Medicine, Department of Human Genetics, Decatur, GA

The Emory Genetics Metabolic Nutrition Program has created a unique initiative called Medical Nutrition Therapy for Prevention (MNT4P) that aligns with the Georgia Department of Public Health and Newborn Screening (NBS) program. MNT4P was established to bridge insurance coverage gaps by providing comprehensive medical nutrition therapy (MNT) to Georgia patients with inherited metabolic disorders primarily identified by NBS. The goal of MNT4P is to provide predictive, preventative, and personalized MNT by facilitating dynamic pathways of nutrition care at diagnosis and throughout the lifecycle. The program aims to be a national model for collecting data, improving quality of life, and innovating healthcare delivery. MNT4P has created a community that connects public health and insurance sectors, consumers, pharmaceutical industry, healthcare providers, and informatics personnel. This allows the program to provide holistic support to patients who are unable to obtain medical foods, low-protein modified foods, dietary supplies, insurance navigation and nutrition education. MNT4P has identified gaps in access, monitoring, and diet adherence, which are often associated with inadequate health insurance coverage. A key component of MNT4P is the opportunity for additional nutrition services including insurance navigation. At just 1.5 years after inception, MNT4P has provided services to approximately half of Emory's active genetic metabolic patient population. This presentation will include data on the identified gaps and how MNT4P has worked to bridge them for patients. The data will demonstrate the benefit of an integrated NBS program that identifies patients and provides immediate and ongoing support in lifelong diet management for improved health outcomes.

A CASE REPORT OF CLASSIC RETT SYNDROME WITH ABSENCE OF *MECP2* MUTATION AND PRESENCE OF *CTNNB1* MUTATION

Shakil S[1,2], Aronoff E [3], Daniel Tarquinio D[1,3]
[1]Center for Rare Neurological Diseases, Atlanta, GA; [2]University of Georgia, Athens, GA; [3]Emory University, Atlanta, GA

Introduction: Rett syndrome (RTT) is a rare neurodevelopmental disorder characterized by developmental regression, stereotypical hand movements, seizures, and movement disorder. Although most individuals with classic RTT have a mutation in the *MECP2* gene, some without the *MECP2* mutation have no identifiable genetic etiology for the syndrome. Rare exceptions exist, however, that are usually associated with atypical RTT, rather than classic RTT. We present an individual who is diagnosed with classic RTT despite absence of a *MECP2* mutation. **Method**: Whole exome sequencing was completed using genomic DNA of a whole blood sample from the patient. EmExome is performed on genomic DNA using Agilent VPlus targeted sequence capture method. The sequence is compared with the published human genome build UCSC hg19 reference. After variants of unknown significance were detected, the parents also received the same sequencing to evaluate which mutation occurred *de novo* in the patient. **Results**: A pathogenic variant of unknown significance was discovered in the β-catenin gene (*CTNNB1)* gene in this patient. This variant was not found in the individual's mother or father indicating that this variant likely occurred *de novo* in this individual. **Conclusion**: *CTNNB1* mutations have been reported to be associated with intellectual disability syndrome, significant motor delay with hypotonia of the trunk and (progressive) distal hypertonia/spasticity of the legs, speech impairment, behavioral anomalies, frequent microcephaly and overlapping facial features. Our patient's phenotype was consistent with these features. In addition, the patient is also diagnosed with classic Rett syndrome despite the absence of a *MECP2* mutation.

POSTER PRESENTATIONS
(Alphabetical by presenting author)

GENETIC COUNSELING VIA TELEMEDICINE IN THE CF NEWBORN SCREEN – A SUCCESSFUL TRIAL AND FUTURE DIRECTIONS

Jonasson A[1], Delgado-Villalta S[2], Zori R[1], Hopfer S[3], Collins M[3], Stalker H[1]
[1]Division of Pediatric Genetics & Metabolism, University of Florida; [2]Division of Pediatric Pulmonary, University of Florida, Gainesville, FL; [3]Connecticut Newborn Screen, University of Connecticut,Storrs, CT

Most NBS programs mandate genetic counseling, but this service can quite variable depending on the Center. Genetic counseling is frequently not provided by genetic counselors (GCs) due to funding constraints or availability of a genetic counselor within the CF treatment centers. There are two major issues that are significant to the provision of better GC services to families who are identified through the CF NBS program: reducing inappropriate anxiety by ensuring that families have appropriate information about the screen, the risks of CF, and the process that will be involved in coming to diagnostic resolution about their child, and ensuring that families have a better understanding of the genetic implications of having a child identified as being a carrier for CF, the possibility of having a future child who has CF, and the screening of other family members that is appropriate given the specific structure of the family and the specific mutations identified. We have provided GC via telemedicine to patients identified as having abnormal newborn screens for CF at four disparate locations across the country: UF, UConn, WVU and the State of Utah. Following provision of GC, families completed a satisfaction survey regarding the quality of the information provided to them, and their comfort with the use of the telemedicine modality for provision of this information. Further evaluation of knowledge about CF based on the Ciske index and an interview that was analyzed for content themes.

FROM NEWBORN SCREENING TO NEWBORN GENOMIC SEQUENCING: COMBINING THE POWER OF WGS WITH CORD BLOOD STORAGE THROUGH THE GENERATION™ PROGRAM

Tanner AK[1], Kraus M[1], Chin E[1], Valencia CA[1], Irzyk G[1], Szekeres E[1], Ma Z[1], Hegde M[1,2]
[1]PerkinElmer Genomics, Pittsburgh, PA; [2]Emory University, Atlanta, GA

Newborn screening (NBS) is a successful public health program that enables early identification of individuals with metabolic, endocrine, and other disorders, creating opportunities for early intervention and reduced morbidity and mortality. NBS is currently conducted mainly through biochemical assays, which are inexpensive and quick to perform, often using molecular methods as second-tier follow-up testing. With the advent of next generation sequencing (NGS), the costs for molecular testing have dramatically decreased, bringing the possibility of using molecular methods as primary NBS testing modalities into the realm of possibility. The most comprehensive molecular test currently available is whole genome sequencing (WGS). Nearly 80 life-threatening diseases can be treated using blood stem cells, including metabolic diseases, cancers, blood diseases, and immune disorders. Cord blood has been shown to be effective in lessening the symptoms of cerebral palsy and research is currently being conducted into its efficacy for treating autism. Currently, however, as many of these disorders are not detected by NBS programs, the need for these therapies is often not known until the optimal treatment windows may have passed. We have implemented the Generation™ Whole Genome Sequencing Test to combine the power of WGS with the potential of cord blood and tissue banking. We have optimized WGS on stored cord blood and dried blood spot (DBS). Data will be presented on results from the first 50 samples. Programming issues, such as consenting and patient education, return of results, and unexpected or unusual outcomes or case studies, will be discussed.

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Phone: 704-450-1933
Representatives: Danielle Hilton
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EXHIBITORS AND SUPPORTERS

Agilis Biotherapeutics
6 Kimball Lane
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Phone: 651-216-2966
Representative: Terry Granec
www.agilisbio.com

Agilis Biotherapeutics is advancing innovative DNA therapeutics designed to provide long-term efficacy for patients with ddebilitating, often fatal, rare genetic diseases that affect the central nervous system.

Alexion
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Representative: Lisa Quirk
www.alexion.com

Alexion is a global biopharmaceutical company focused on serving patients and families affected by rare diseases through the innovation, development and commercialization of life-changing therapies.

Alexion Pharmaceuticals
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Representatives: P J Price and Jodie Diamond
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Cambrooke Therapeutics
4 Copeland Drive
Ayer, MA 01432
Phone: 630-660-4930
Representative: Dan Kilbride
www.Cambrooke.com

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EXHIBITORS AND SUPPORTERS

Cycle Pharmaceuticals, Ltd.
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Representatives: Chris Abbott, Brian Sergeian
www.cyclepharma.com

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Phone: 800-309-1673
Representative: Catherine Allen
www.GGC.org

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Nashville, TN 37208
Phone: 615-587-3766
Representative: Leslie Allen
www.invitae.com

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EXHIBITORS AND SUPPORTERS

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P.O. Box 110409
Research Triangle Park, NC 27709
Phone: 910-616-6341
Representative: Axel Kusber
www.metabolon.com

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Representative: Matt Sergent
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Representative: Robert Menendez
www.preventiongenetics.com

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Representatives: Kevin Fowler and John King
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Representative: Shawn Hayes
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SERN at Emory University
100 Woodruff Circle, 7th Fl, Suite 7130
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Phone: 404-778-8521
Representative: Saran Gurung
www.southeastgenetics.org

The Southeast Regional Network (SERN) at Emory University is a HRSA-funded initiative that strives to improve health equity and health outcomes in individuals with genetic conditions, reduce morbidity and mortality caused by genetic conditions (including congenital and metabolic disorders); and to improve the quality of coordinated and comprehensive genetic services to children and their families.

The Smith Family Clinic for Genomic Medicine
701 McMillian Way NW, Suite A
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Phone: 256-327-9640
Representative: Carol Aiken
www.smithfamilyclinic.org

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Representative: Kimberly Phelps-Weber
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Ultragenyx is a clinical-stage biopharmaceutical company committed to bringing to market novel products for the treatment of rare and ultra-rare diseases, with a focus on serious, debilitating genetic diseases.

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Columbia, SC

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Huntsville, AL

Catherine Allen, MS, CGC
Greenwood, SC

Leslie Allen
Nashville, TN

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Asheville, NC

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New Orleans, LA

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Alexa Bledsoe, MS
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Megan Boothe, MD
Birmingham, AL

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Jon Bos, PsyD
Columbia, SC

Beth Boyea, MS
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Candice Brannen, PhD
Apex, NC

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Waycross, GA

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New Orleans, LA

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Dick Buchanan
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Julie Cheek, EDS
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Clemson, SC

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Knoxville, TN

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Katy Drazba, MPH, MS, CGC
Florence, SC

Barb DuPont, PhD
Greenwood, SC

Beth Dworetsky, MS
Albuquerque, NM

Dusty Edwards, BSN, RN
Tallahassee, FL

Armand Evangelista
Lake Forest, IL

Stephanie Evenson, BSN
Alpharetta, GA

PRE-REGISTERED ATTENDEES

Meagan Farmer, MS, CGC
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Ben Filas, PhD
Waltham, MA

Venessa Fonte
Farmingdale, NY

Shandrea Foster, MS
Spartanburg, SC

Kevin Fowler, BSBA
Elmhurst, IL

Mike Garofalo, BS
Farmingdale, NY

Jonathan Gattis, BA
Waxhaw, NC

Matthew German
Lake Forest, IL

Terry Granec
Lynnfield, MA

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Raleigh, NC

Saran Gurung
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Miami, FL

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Decatur, GA

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Novato, CA

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Gretna, LA

Danielle Hilton
Mooresville, NC

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Mandeville, LA

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McKinney, TX

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Anna Hurst, MD
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Natalye Jones, BSN
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Atlanta, GA

Robert Latour, PhD
Clemson, SC

Mandy Ledford
Lexington, MA

Jennifer Lee, PhD
Greenwood, SC

Stacey Lee, PhD
Durham, NC

Yuwen Li, MD, PhD
New Orleans, LA

PRE-REGISTERED ATTENDEES

Timothy Lim, PhD
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Charlotte, NC

Michael Lyons, MD
North Charleston, SC

Jerry Main
Lebanon, NJ

Priyanshu Mathur, MD
Atlanta, GA

Molly McDevitt, RD
Nashville, TN

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Yetsa Osara, MPH
Atlanta, GA

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Charlotte, NC

Andrea Paal
Marlborough, MA

Ashley Park, MS
Atlanta, GA

Bharat Patel, PharmD
Stanford, FL

Ravi Pathak, PhD
Lexington, MA

Kostas Petritis, PhD
Atlanta, GA

Kimberly Phelps-Weber
Navato, CA

Billy Philips, PhD, MPH
Lubbock, TX

Beryl Polk, PhD
Jackson, MS

Laura Pollard, PhD
Greenwood, SC

Amy Ponte, PhD
Beaufort, SC

Marty Porter, PhD
Decatur, GA

Cynthia Powell, MD
Chapel Hill, NC

Belkys Prado, RD, LDN
Tampa, FL

Graeme Preston, BA
New Orleans, LA

PJ Price
New Haven, CT

Theresa Pringle, MPH
Atlanta, GA

Emily Prizer, BS
Alexandria, VA

Lisa Quirk
New Haven, CT

Melissa Rich, BS
Port Orange, FL

Kacie Riley, BA MS
Durham, NC

Amy Robertson, MSN
Nashville, TN

Ami Rosen, MS, CGC
Atlanta, GA

Suleima Salgado, MBA
Atlanta, GA

Mary Lauren Salvatore, MPH
Atlanta, GA

Rossana Sanchez Russo, MD
Atlanta, GA

Sarah Savage, MS, CGC
Boston, MA

Joseph Schneider, PharmD
Woodbury, MN

Brian Sergenian, BASc
Boston, MA

Matt Sergent
Tarrytown, NY

PRE-REGISTERED ATTENDEES

Sidra Shakil, BS
Norcross, GA

J Daniel Sharer, PhD
Birmingham, AL

Scott Shone, PhD
Research Triangle PK, NC

Jill Shuger, ScM
Rockville, MD

Mary Rose Simpson, BS
Lancaster, SC

Rani Singh, PhD, RD
Atlanta, GA

Lloyd Sirmons, BS
Blackhear, GA

Jana Smith
Marlborough, MA

Susan Sparks, MD, PhD
Cambridge, MA

Charles Spaulding
Novato, CA

Catherine Spellicy, PhD
Asheville, NC

Tanya Spells, MS
Columbia, SC

Banning Stack, MS
Signal Mountain, TN

Jessica Stack, MArch
Signal Mountain, TN

Linda Starnes, BS
Longwood, FL

Cheryl Steep, MS, RD, LD
Duluth, GA

Erica Stelten, RD
Decatur, GA

Adrya Stembridge, AS
Atlanta, GA

Fabiola Storz
Novato, CA

Cindy Strange, MBA
Orlando, FL

Marshall Summar, MD
Washington, DC

Thomas Terrell, MD
Clinton, TN

Missy Tharp, MN
Jackson, MS

Helen Travers, MS
Miami Shores, FL

Julie Tucker, BA
Harvest, AL

Jeff Turnbull
Santa Clara, CA

Nikky Ugwuoke, PharmD
Waxhaw, NC

Alexander Valencia, PhD
Branford, CT

Elizabeth Vengoechea, MS
Atlanta, GA

Ashley Volz, MS, CGC
New Orleans, LA

Brianna Volz
Aliso Viejo, CA

Bryan Voss, PhD
Nashville, TN

Thuy Vu, MS
Winston-Salem, NC

Angela Walter, MS, CGC
Hillsborough, NC

Michael Watson, PhD
Bethesda, MD

Lance Webb, BS
The Woodlands, TX

Brandon Welch, PhD
Johns Island, SC

Jill White, LCSW
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Tim Wood, PhD
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