2017 MEETING LOCATIONS

DATE/TIME	FUNCTION	LOCATION
Thursday, July 20		
8:30 a – 10:00 a	Public Health Session: LTFU Workgroup	Victoria
10:00 a – 12:00 p	Telegenetics Workgroup	Alexander
12:00 p – 1:00 p	ISS-Sanofi Genzyme	Swannanoa
12:00 p – 5:00 p	Registration	Salon A Foyer
1:00 p – 6:00 p	SERN Platform Session 1	Salon A
3:20 p – 3:50 p	Break	Salon A Foyer
6:30 p – 8:30 p	Reception/Poster Session	Salon B-C
Friday, July 15		
7:30 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	SERGG Platform Session 2	Salon A
10:15 a – 10:45 a	Break	Salon B-C
10:45 a – 11:45 a	SERGG Platform Session 3	Salon A
12:00 p – 1:00 p	Lunch	Windsor Ballroom and Foyer
1:00 p – 2:45 p	SERGG Platform Session 4	Salon A
2:45 p – 3:00 p	Travel to Concurrent ISS	
3:00 p – 5:15 p	Concurrent Industry-Supported Symposium	
3:00 p – 5:15 p	Horizon Pharma	Victoria
3:00 p – 4:00 p	GeneDX	Swannanoa
3:00 p – 4:00 p	Shire	Berkeley
4:00 p – 4:15 p	Break	
4:15 p – 5:15 p	PerkinElmer	Swannanoa
4:15 p – 5:15 p	Shire	Berkeley
CONCURRENT SESSION – CONSUMER ALLIANCE		
8:30 a – 10:15 a	Consumer Alliance Plenary	Alexander
10:15 a – 10:45 a	Break	Alexander
10:45 a – 12:00 p	Consumer Alliance Plenary	Alexander
12:00 p – 1:00 p	Lunch	Winsor Ballroom and Foyer
1:00 p – 2:00 p	Consumer Alliance Plenary	Alexander
2:00 p – 2:15 p	Break	Alexander
2:15 p – 3:00 p	Consumer Alliance Plenary	Alexander
3:00 p – 5:15 p	Concurrent Industry-Supported Symposium	Various Locations (See above)
Saturday, July 16		
7:00 a – 8:00 a	Continental Breakfast	Salon B-C
7:00 a – 10:30 a	Exhibits/Posters	Salon B-C
8:00 a – 10:30 p	Registration Desk Open	Salon A Foyer
8:00 a – 9:45 a	SERGG Platform Session 5	Salon A
9:45 a – 10:15 a	Break	Salon B-C
10:15 a – 12:00 p	SERGG Platform Session 6	Salon A
12:00 p – 12:30 p	SERGG Business Meeting	Salon A
CONCURRENT SESSION – CONSUMER ALLIANCE		
8:00 a – 10:00 a	Consumer Alliance Plenary	Swannanoa
10:00 a – 10:15 a	Break	Swannanoa
10:15 a – 12:00 p	Consumer Alliance Plenary	Swannanoa
12:00 p – 12:30 p	SERGG Business Meeting	Salon A

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

PLATFORM PRESENTATIONS
(In Order Presented)

THE NC NEXUS PROJECT OF NEWBORN EXOME SEQUENCING

Powell CM[1]; Roche MI[1]; Rini C[2]; Lewis MA[3]; Paquin RS[3]; Bailey DB[3]; Margolis M[1], Butterfield R[1]; Milko L[1]; Powell B[1]; Berg JS[1]
[1]University of North Carolina at Chapel Hill, [2]Hackensack University Medical Center, [3]RTI International

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 being recruited: children from birth to age 5 years with known conditions diagnosed through standard newborn screening with disorders such as inborn errors of metabolism or hearing loss and a second consisting 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) based on expert opinion utilizing a semi-quantitative metric scoring system. Families in each cohort will be randomized; those in the "Decision" group have an option to receive additional results of pathogenic variants in genes associated with conditions that have childhood onset but are not medically actionable, that have adult onset and medical actionability, or are associated with carrier status. Results: Since recruitment began in June 2016 to date, 186 patients have been recruited into the project and 24% agreed to have their child's genome sequenced. Parents must complete an on-line decision aid to participate and of those who do, approximately 70% agreed to have their child's genome sequenced. Reasons given by active decliners include concern for child, too busy, too far to travel, not interested, do not want additional testing and do not want to share information. Recruitment is ongoing and updated data will be reported.

FOCUSED EXOMES: A TARGETED GENOMIC TEST WITH A SAFETY NET

Bend EG, Louie RJ, Bend RC, McGee S, Kubiak K, Lee JA, Friez MJ, Jones JR
Greenwood Genetic Center

The Greenwood Genetic Center is currently offering a novel type of next generation sequencing (NGS) assay that we term Focused Exome Sequencing (FES). FES samples are processed using our standard whole exome sequencing (WES) bench pipeline, but only 1 to 20 physician-requested genes are analyzed. This targeted approach may offer key benefits over WES and fixed NGS panels for a subset of patients. First, it allows for customized genotyping specific to a patient's unique phenotype, and therefore the diagnostic yield of this test is relatively high. We reported a pathogenic or likely pathogenic variant in over 50% of FES cases (n=36) since this test was launched. This is significantly higher than the yield for WES (36%, n=413) or that of our high-yield panels (e.g. Skeletal Dysplasia, 20%, n=131). Second, FES can potentially target genes for which clinical Sanger sequencing does not currently exist, increasing the diagnostic options for patients. Third, data analysis and reporting are more straightforward in comparison to both WES and panels. This has the potential of easing the burden on the laboratory, the clinicians and the patients, since no variants of uncertain significance in unrelated genes will be reported. Finally, in the event of a negative result by FES, reflexing to WES analysis is possible because the enrichment method is the same, thus providing an important diagnostic safety net for our patients. Here we will outline our methodology and results, and highlight interesting cases we have solved by FES.

PLATFORM PRESENTATIONS
(In Order Presented)

SUSCEPTIBILITY TO POST-TRAUMATIC STRESS DISORDER (PTSD) IS COMORBID WITH MITOCHONDRIAL DYSFUNCTION IN MICE

Preston G[1], Emmerzaal T[2], Kirdar F[1], Morava E[1] and Kozicz T[1]
[1]Tulane University School of Medicine; [2]Radboud University Medical Center, Nijmegen, Netherlands

Psychopathology is frequently associated with mitochondrial dysfunction in patients. Post-traumatic stress disorder (PTSD) is a debilitating psychiatric disorder that affects millions of people worldwide. Induced by exposure to a traumatic event, the duration and symptomatology of the disorder is highly heterogenous, and the severity of the inducing trauma does not predict the severity of the disorder, suggesting the possibility of an underlying genetic metabolic factor. We investigated whether mitochondrial dysfunction is associated with susceptibility to PTSD. We exposed 48 wildtype male mice of the FVB.129P2 background to a PTSD-induction paradigm previously shown to reliably induce PTSD-like symptomatology with a similar presentation to that of PTSD in humans. The inducing trauma was inescapable electric foot shock, administered in two decontextualized sessions over two consecutive days. PTSD-susceptible animals were diagnosed through a series of behavioral tests to identify physical symptoms of PTSD. 12 animals most and least clearly displaying PTSD-like symptomatology were identified, brain tissue was collected, and mitochondria were isolated. The activities of the electron transport chain (ETC) complexes I through IV, as well as citrate synthase, were measured to determine mitochondrial activity and density, respectively. Comparing ETC activity and mitochondrial density to PTSD-symptomatology reveals a statistically-significant inverse relationship between PTSD susceptibility and mitochondrial capacity ($p=0.016$). Our results indicate that susceptibility to PTSD in mice is comorbid with mitochondrial dysfunction, suggesting the possibility that mitochondrial dysfunction may be sufficient to induce PTSD susceptibility in mice. Future research directions include exacerbating mitochondrial dysfunction and assaying for a concordant increase in PTSD susceptibility.

COPY NUMBER VARIANT ANALYSIS ENHANCES MOLECULAR DIAGNOSTIC YIELD OF INBORN ERRORS OF METABOLISM

Ho Y-Y, Truty R, Murillo L, Beltran D, Harte R, Morgan C, Fuller A, White H, Fox M, and Johnson B
Invitae Corp, San Francisco, CA

Inborn errors of metabolism (IEM) are identified in an estimated 1 in 5000 live births. Newborn screening programs (NBS), which are among the most successful public health programs, enable early recognition and management of IEM and prevent IEM-associated morbidity, disabilities, and death. Molecular confirmation of pathogenic mutations ascertains a diagnosis for individuals suspected of having IEM and further justifies evidence-based treatment and medical interventions. In this study, DNA samples from 352 individuals suspected with IEM by either positive NBS or clinical evaluation were analyzed with next-generation sequencing. Variants were classified using an evidence-based variant interpretation system that conforms to the ACMG guidelines. Comprehensive analysis of 162 genes, including metabolic genes, followed by variant classification showed that copy number variants (CNVs) composed 10% of the likely pathogenic (LP) or pathogenic (P) mutations. In the 63 NBS metabolic genes subgroup, CNVs composed 5% of the LP/P variants, and the LP/P CNVs were enriched in the ACADM and PRODH genes. Notably, ACADM is the causal gene for medium chain acyl-CoA dehydrogenase deficiency (MCAD), the most common autosomal recessive IEM. CNVs account for 27% of unique LP/P pathogenic variants identified in ACADM. In addition, novel CNV mutations were identified in the ACADM and SLC35A2 (causes congenital disorder of glycosylation, type IIm) genes. In conclusion, CNV detection expands the mutation spectrum and improves the molecular diagnostic yield of IEM. Therefore CNV analysis should be included as a routine practice for the diagnosis and confirmation of IEM.

PLATFORM PRESENTATIONS
(In Order Presented)

METABOLIC PATHOGNOMONICS: INCORPORATING DISEASE-SPECIFIC BIOCHEMICAL DATA IMPROVES VARIANT INTERPRETATION FOR INHERITED METABOLIC DISORDERS

Harte R, Murillo L, Beltran D, Fox M, Fuller A, Ho Y-Y, Morgan C, White H, Topper S, Nykamp K[1], Johnson B
Invitae, San Francisco, CA, USA

Commercial laboratories routinely use patient clinical information provided by ordering physicians so that, when appropriate, clinical symptoms specific to a patient's phenotype can be used as supporting evidence for variant classification. The American College of Medical Genetics and Genomics (ACMG) variant interpretation guidelines reflect this practice but are not specific about its implementation. To augment this guideline, we developed a new category of evidence and incorporated it into Sherloc, our points-based system for variant interpretation. These new categories of evidence provide more detailed criteria combining distinctive phenotypic data with specific gene-level information. Inherited metabolic disorders are unique in that the phenotypes of affected individuals include biochemical information that is highly specific to the condition and, in many cases, diagnostic. Therefore, we incorporate results from clinical biochemical testing by granting points during the interpretation process. In addition, when published literature shows that the diagnostic yield for a disorder is >75% when patients have specific biochemical results, we consider these results as pathognomonic. Variants in cases that meet these more stringent biochemical criteria and have the expected genotype are weighted with additional points toward a pathogenic classification. For autosomal recessive inherited metabolic disorders, we combine this systematic method of assessing phenotypic data with variant phasing information, which provides a powerful approach for the interpretation of novel variants. As of March 2017, 72% unique variants of those eligible for the application of these criteria have been classified as likely pathogenic or pathogenic for multiple metabolic disorders (e.g., homocystinuria, citrullinemia).

ONE YEAR FOLLOW-UP OF B VITAMIN AND IRON STATUS IN PATIENTS WITH PHENYLKETONURIA RESPONSIVE AND NON-RESPONSIVE TO SAPROPTERIN

Brantley K[1], Douglas TD[2], and Singh RH[2]
[1]Emory University Rollins School of Public Health; [2]Emory University School of Medicine, Department of Human Genetics, Metabolic Nutrition Program

Background: People with Phenylketonuria (PKU) who respond to sapropterin (tetrahydrobiopterin-BH_4) with lower blood Phe are often able to increase phenylalanine (Phe) tolerance and/or decrease medical food while maintaining therapeutic blood Phe concentrations. B12, B6, folate, and iron intake are concerning due to higher presence in PKU medical foods that may be reduced in sapropterin responsive patients while not adequately compensated by increases in intact foods. **Methods**: Diet and blood nutrient outcomes of sapropterin responders and non-responders at baseline and one year (n=33) were measured. Subjects were males and females, age 4-50 years. Blood markers for iron, B12, B6, and folate were analyzed by Quest Diagnostics. Assessment of 3-day diet records completed with NDSR by a registered certified dietitian. Patients were matched to NHANES controls for further laboratory comparisons. Chi-square, t-tests, and linear regression (alpha=0.05) were performed using SAS software (version 9.4). **Results**: Diet intake associated with lab values for folate (p=0.029), but not for other nutrients, once controlled for age. Both response groups had stable dietary intake of examined nutrients, maintained labs within reference ranges, and met DRI guidelines across one year. However, responders' serum B12 declined significantly by one year (p=0.014). Additionally, serum B12 in both response groups was lower than NHANES controls at both study visits. Labs for B6, folate, and iron did not differ from controls. **Conclusion**: Although mean dietary and lab values for B12, B6, folate, and iron remained within reference limits in PKU patients regardless of sapropterin, serum B12 significantly declined in responders over one year. Lab values corresponded with diet intake for folate only. Although dietary or lab measured iron, B12, B6, and folate deficiencies were minimal in our study cohort, assessing micronutrient status in sapropterin responsive PKU remains pertinent.

PLATFORM PRESENTATIONS
(In Order Presented)

AT-HOME URINE TEST FOR BLOOD PHE-LEVEL MONITORING FOR PHENYLKETONURIA (PKU)

Latour RA[1], Chumanov G[2], Whitehead DC[2], Narangoda C[2], DeLuca JM[3,4], Wood T[4], and Champaigne NL[4]

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Phenylketonuria (PKU) is a rare metabolic disorder caused by a genetic defect in the liver enzyme that converts phenylalanine (Phe) to tyrosine. Untreated, elevated Phe levels are neurotoxic and lead to progressive, irreversible intellectual impairments within a few months after birth. Treatment for PKU is effective in preventing the neurological complications, but involves strict metabolic control using a low-Phe diet supplemented with specialized medical foods combined with close monitoring of blood Phe levels to achieve a targeted range of 2-6 mg/dL. Currently, Phe levels are typically monitored by mailing dried blood spots to a clinical laboratory for analysis, which takes 5-10 days for results to be received, leaving patients susceptible to unrecognized spikes in Phe levels while waiting for results. To address this problem, we have been working on the development of a simple at-home urine test for blood Phe-level monitoring. Our approach uses colorimetric test coupons that detect phenylpyruvic acid (PPA) and creatinine (CRE) concentration in urine. After exposure to urine, the coupons are photographed in a light-controlled environment using a smartphone and analyzed using an RGB color app to determine PPA and CRE concentrations. The ratio of PPA/CRE is then used to assess the blood Phe level. Our preliminary results from over 10 PKU patients show strong correlation between urine PPA/CRE ratios and blood Phe levels for blood-Phe levels over a range of 1-21 mg/dL, thus demonstrating this novel urine test as a potential low-cost, simple, noninvasive method for at-home blood Phe-level monitoring for the PKU community.

RECOGNITION OF NEUROFIBROMATOSIS TYPE 1 WITH FACIAL DYSMORPHOLOGY NOVEL ANALYSIS

Cignetti CA[1], Hurst ACE[2]

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A wide variety of dysmorphic facial features have been reported in people with the genetic condition Neurofibromatosis type 1 (NF1), but a characteristic facial gestalt has not been described. NF1 has not previously been studied with facial analysis software. The primary objective of this study is to determine whether NF1 can be recognized by a distinct facial pattern, and to assess for *NF1* genotype-facial phenotype relationships. To date, photographs of 29 individuals with clinical and/or molecular diagnoses of NF1 have been captured and analyzed by Facial Dysmorphology Novel Analysis (FDNA) software, a technology that generates topographical data from two-dimensional images. Recruitment is ongoing. Data is assessed for patterns within the NF1 cohort, associations between *NF1* genotype and facial phenotype, and relationships to other genetic conditions previously characterized by FDNA. Additional studies are underway to compare this cohort to individuals with related genetic disorders including Noonan syndrome and other RASopathies. The facial pattern associated with NF1 will be added to the FDNA database, and the NF1 facial gestalt will be screened for with every FDNA search. This may support an early diagnosis of NF1, and the non-invasive, cost-free FDNA technology could be a screening resource for this condition. Benefits include saving time and money that might be spent exploring other causes of symptoms, and knowing to screen for and manage potentially life-threatening complications associated with NF1.

PLATFORM PRESENTATIONS
(In Order Presented)

X CHROMOSOME DELETION ASSOCIATED WITH MOSAICISM IN TWO GENERATIONS OF FEMALES

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Follow-up testing for positive noninvasive prenatal testing (NIPT) results has, at times, revealed unexpected maternal and/or fetal karyotypes. In one such case a 31 year old G4P3 woman's NIPT result indicated an increased risk for monosomy X in her female fetus. Prenatal ultrasound was normal. The baby was born at 39 weeks gestation with normal growth and physical appearance. Analysis of the child's peripheral blood chromosomes showed 46,X,del(X)(q27) in 46/50 cells. Two of 50 cells were missing the deleted X (45,X) pattern, and 2 cells had 47,X,del(X)(q27),del(X)(q27). Analysis of the mother's chromosomes yielded similar results (3 cell lines). Interphase FISH studies for each individual supported the presence of cell lines with loss and gain of an X. Microarray analysis of the mother's blood indicated a 9.8 Mb terminal deletion beginning at Xq27.3 without additional significant copy variations. At age 6 weeks the proband's physical exam by a geneticist was normal. The mother, although fertile and of normal stature and development, has a history of irregular menses. The maternal grandmother is 4'11", and she has 1 sister with infertility and 1 with repeated pregnancy loss. Similar or larger Xq deletions, occasionally with 45,X mosaicism, have been reported. However, this appears to be a rare family in which mitotic nondisjunction occurred in conjunction with inheritance of an Xq deletion in 2 generations of females. Possible factors contributing to this phenomenon, clinical implications of this finding, and the importance of thorough chromosome analysis after a positive NIPT result will be discussed.

USING ALL OF THE TOOLS IN THE DIAGNOSTIC TOOLBOX FOR LYSOSOMAL STORAGE DISEASES

Pollard L, Huang R, Shouse C, Haley J, Hallman J, Cason A, Zipprer D, Carra D, Propes C, and Wood T
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Historically, the diagnosis of a lysosomal storage disease (LSD) began with clinical suspicion, followed by urine screening and/or enzyme analysis, and then molecular testing to confirm the diagnosis. In the era of newborn screening and whole exome sequencing (WES), this testing algorithm is changing. Patients are being identified pre-symptomatically via newborn screening, and WES or next generation sequencing panels can be used as first-tier tests to identify LSD patients with non-specific clinical phenotypes. Perhaps now, more than ever, it is important to combine clinical, biomarker, enzyme and molecular data to achieve the proper diagnosis. We present multiple clinical cases in which an integrated diagnostic approach was needed. Leukocyte enzyme analysis, urine glycosaminoglycan analysis and *IDUA* gene sequencing together distinguishes between alpha-iduronidase pseudo-deficiency and Mucopolysaccharidosis type I in infants with abnormal newborn screens for MPS I. Biomarker analysis helped distinguish between carrier and affected status in an infant with an abnormal newborn screen for Gaucher disease, reduced beta-glucosidase activity, and compound heterozygosity for a pathogenic variant and a VOUS. Biomarker analysis also helped confirm a diagnosis of ultra-rare beta-mannosidosis in a patient with reduced beta-mannosidase activity and compound heterozygosity for a pathogenic variant and a VOUS. Biomarker and enzyme analysis identified a patient with Morquio syndrome with no identifiable *GALNS* mutations. When next generation sequencing identifies a VOUS in a LSD gene, enzyme and/or biomarker analysis can be used to determine pathogenicity. It is imperative to use all of the tools in our diagnostic toolbox when confronting a potential LSD case.

PLATFORM PRESENTATIONS
(In Order Presented)

ATP1A3 GENE ALTERATIONS IN PATIENTS WITH NEUROLOGIC FEATURES AND ABNORMAL MUSCLE TONE

Baggett LM, Rogers RC, Everman DB, Jones JR, Louie RJ, Deluca JL, Worthington JD, and Champaigne NL
Greenwood Genetic Center

Individuals with alternating hemiplegia of childhood and rapid onset dystonia are frequently identified as having pathogenic alterations in the *ATP1A3* gene; however, the frequency in individuals with a mixed neurological course is less clear. The Greenwood Genetic Center Molecular Diagnostic Laboratory has performed whole exome sequencing (WES) analysis on over 400 probands, with pathogenic *ATP1A3* alterations identified in 5 families (1.2% of families tested), making it the gene with the greatest number of pathogenic alterations identified by WES in our lab thus far. While the genetic findings were ultimately consistent with the clinical histories, *ATP1A3*-related disorders were not previously suspected. We have identified *ATP1A3* gene alterations in one individual with CAPOS syndrome (cerebellar ataxia, areflexia, pectus excavatum, optic atrophy, and sensorineural hearing loss); one individual previously suspected to have a mitochondrial depletion syndrome; one individual with confirmed cystic fibrosis and hypotonia and seizures; 3 siblings with microcephaly, hypotonia, and developmental delay (2 with cerebellar atrophy and seizures); and one individual with nystagmus and dystonia. While all of these patients were seen for a variety of neurologic features, all with some level of muscle tone concerns, they have clinically heterogeneous findings. While most *ATP1A3* pathogenic findings are *de novo*, parental germline mosaicism has been reported and is likely present in one family described here. This is an important consideration when discussing recurrence risks. These results highlight the importance of WES and the wide spectrum of phenotypes associated with *ATP1A3* alterations.

A NEUROPSYCHOLOGIC MODEL OF MELAS DISEASE USING INDUCABLE PLURIPOTENT STREM CELL DERIVED EXCITATORY NEURONS

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Most individuals diagnosed with the common mitochondrial phenotype of MELAS carry the pathogenic variant 3243A>G in their mitochondrial DNA (mtDNA). Patients show a broad range of symptoms, involving mainly the skeletal muscle and the central nervous system. The mutation being present in the maternally inherited mtDNA, makes generating a knockdown/knockout animal models problematic. In order to increase our understanding on the neuropathobiology of mt.3243A>G we created human cortical neurons (iNeurons), derived from inducible pluripotent stem cells (IPSCs) and studied the impact of mt.3243A>G on mitochondrial- and neuronal (including synaptic) function. Patient-derived fibroblasts were reprogrammed to IPSCs, which were tested for heteroplasmy by sanger sequencing; three clones from the same patient, with different heteroplasmy levels (0%, 72%, and 83%) were expanded. These IPSC's were differentiated into excitatory iNeurons by lentiviral rtta- and Neurogenin 2 (Ngn2) expression. Here we report on the characterization of the iNeurons both at the single-cell and neuronal network level. We also describe whole-cell patch clamp recordings assessing spontaneous excitatory post-synaptic currents (sEPSC's), as well as intrinsic properties, combined with Mitotracker, MAP2- and Synapsin immunocytochemical staining. Additionally, we report on multi-electrode arrays (MEA's) used to study the effects of the 3245G>G mutation on the network activity of the iNeurons. Our study illustrates the relevance of our patient-specific in vitro neurophysiologic model of mitochondrial involvement, which will enhance our understanding of the role of mitochondrial dysfunctions in neurological manifestations of MELAS disease.

PLATFORM PRESENTATIONS
(In Order Presented)

THE HEART OF THE MATTER: CARDIAC INVOLVEMENT IN PATIENTS WITH LATE-ONSET POMPE DISEASE AND THE C.-32-13T>G VARIANT

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Background: The "leaky "c.-32-13T>G splice site variant is the most common pathogenic variant in Caucasian patients with late-onset Pompe Disease (LOPD). Characterization of potential cardiac manifestations, especially severe manifestations such as hypertrophic or dilated cardiomyopathy (HCM, DCM), will aid in the development of clinical management guidelines for increasing numbers of patients identified through newborn screening (NBS). **Objective:** To describe cardiac manifestations associated with the c.-32-13T>G variant and, **Methods:** 1) medical record review of the Duke Pompe LOPD cohort heterozygous for c.-32-13T>G [n=83 (61%), median age 48 years], 2) review of NBS data from three states and, 3) literature review. **Results**: Twenty-six (33%) patients from the Duke cohort had some cardiac involvement; LVH, LA enlargement, mild valvular lesions; and arrhythmias such as 1st degree A-V block, RBBB, and prolonged QTc. LVH may be attributable to comorbidities such as hypertension, diabetes or hyperlipidemia seen in these patients. NBS in IL, MO and NY States identified 54 children with the c.-32-13T>G variant; none had cardiac involvement on baseline ECG/ECHO or on follow-up. HCM and DCM were absent in the Duke cohort and the NBS cohort, which is consistent with reports in the literature. **Conclusions:** While LOPD patients with the c.-32-13T>G variant may develop mild cardiac manifestations over time, there is no evidence that this variant is associated with HCM or DCM. Children diagnosed with LOPD through NBS who are heterozygous for the c.-32-13T>G variant can be followed with ECG/ECHO yearly, thereby reducing healthcare costs and anxiety in patients and families.

ORAL D-GALACTOSE SUPPLEMENTATION IN PGM1-CDG

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Background/Purpose: Phosphoglucomutase-1 deficiency is a subtype of congenital disorders of glycosylation (PGM1-CDG). Previous case-reports in PGM1-CDG patients receiving oral D-galactose (D-gal) showed clinical improvement. So far no systematic *in vitro* and clinical studies assessed safety and benefits of D-gal supplementation. In a prospective pilot study, we evaluated the effects of oral D-gal in nine patients. **Methods/Results:** D-gal supplementation was increased to 1.5 g/kg/day (maximum 50 g/day) in three increments over 18 weeks. Laboratory studies were performed before and during treatment to monitor safety and effect on serum transferrin-glycosylation, coagulation, liver and endocrine function. Additionally, the effect of D-gal on cellular glycosylation was characterized *in vitro*. Eight patients were compliant with D-gal supplementation. No adverse effects were reported. Abnormal baseline results (ALT/AST/aPTT) improved or normalized already using 1g/kg/day D-gal. Antithrombin-III levels and Transferrin-glycosylation showed significant improvement, and increase in galactosylation and whole glycan content. *In vitro* studies before treatment showed N-glycan hyposialylation, altered O-linked glycans, abnormal LLO-profile, and abnormal nucleotide-sugars in patient fibroblasts. Most cellular abnormalities improved or normalized following D-gal treatment. **Conclusion:** D-gal increased both UDP-Glc and UDP-gal levels and improved LLO fractions in concert with improved glycosylation in PGM1-CDG. Oral D-gal supplementation is a safe and effective treatment for PGM1-CDG in this pilot study. Transferrin glycosylation and ATIII levels were useful trial end points. Larger, longer duration trials are ongoing.

PLATFORM PRESENTATIONS
(In Order Presented)

IMPLEMENTATION OF NEWBORN SCREENING FOR POMPE DISEASE, MPS-I AND X-LINKED ADRENOLEUKODYSTROPHY IN GEORGIA

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Emory University, EGL Genetic Diagnostics, and the Georgia Department of Public Health were jointly awarded funding to perform pilot implementation projects for newly recommended disorders added to the newborn Recommended Uniform Screening Panel including Pompe disease (PD), mucopolysaccharidosis type I (MPS-I) and X-linked adrenoleukodystrophy (XALD). Testing for MPSI and PD began with an initial two-plex enzyme assay using tandem mass spectrometry then interpreted with the assistance of post-analytical tools from the Collaborative Laboratory Interpretive Reports (CLIR). Positive results trigger second tier testing via a 6-plex enzyme assay. If the second tier assay is positive for any screened condition, the infant's provider is contacted and follow-up procedures are initiated. The testing process is similar for XALD; however, second tier testing is performed via a chromatography assay. At the time of submission, over 18,000 samples have been tested for Pompe and MPS-I, with 1.5 – 2% referred for 2nd tier testing. The referral rate for second tier testing for X-ALD has not yet been determined. The analysis using post-analytical tools allows for comparison of patient samples with both Georgia specific references ranges and ranges from laboratories across the country. In addition, the tools may help differentiate true positives, false positives, and carriers. Combined, this reduces the burden on follow-up programs and families and improves the overall performance of the screening program.

GREENWOOD GENETIC CENTER TELEHEALTH INITIATIVE

Lyons MJ, Lynch JB, and Schwartz CE
Greenwood Genetic Center

The Greenwood Genetic Center (GGC) is a nonprofit organization founded in 1974 to serve as a resource for all individuals who need genetic services or information and to reduce the prevalence and impact of genetic disorders. GGC functions as a resource to residents of South Carolina at our home campus in Greenwood and satellite offices in Charleston, Columbia, Florence and Greenville. GGC has developed a telehealth initiative in order to further ensure that all individuals and families in South Carolina have access to genetic services. Our initial efforts are focusing on utilizing telehealth at our Florence office as there is decreased access to genetics care in Florence due to the lack of a clinical geneticist in the Florence area. Clinical geneticists from the GGC Charleston office started using telehealth in November 2016 to evaluate patients due for follow-up appointments in Florence. The use of telehealth has increased the number of available appointments for patients in the Florence office. As we gain experience, we anticipate offering additional telehealth appointment slots for follow-up appointments as well as new referrals. In order to monitor the success of our telehealth initiative, surveys are being used to determine patient and provider satisfaction. We are also collecting data to identify improvements in access to care, travel time, wait time, and cost savings related to telehealth. Results for our initial telehealth initiative in Florence will be presented along with plans to expand our program across all GGC offices to increase access to genetic services in South Carolina.

PLATFORM PRESENTATIONS
(In Order Presented)

URINARY GB_3 IS A USEFUL BIOMARKER FOR MONITORING TREATMENT IN FABRY DISEASE

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Fabry disease is an X-linked lysosomal storage disease characterized by the accumulation of globotriaosylceramide (Gb_3) in tissues. Treatment in the form of ERT (agalsidase beta) has been available in the USA since 2003. Clearance of Gb_3 from tissues in response to ERT correlates with reduced urinary Gb_3. Between 2009 and 2012 a manufacturing shortage of ERT resulted in dose reduction or delay in treatment which provided an opportunity to evaluate the impact of treatment and dosing on urinary Gb_3 levels in Fabry disease. First morning urine was donated by patients with Fabry disease over two years. The samples collected in year 1 occurred during the enzyme shortage, and the samples collected in year 2 were obtained ~6 months after the shortage ended. In comparison to samples collected after the shortage ended: More than half of all subjects had elevated urinary Gb_3; Males had significantly higher concentrations of urinary Gb_3 than females; and 75% of treated males had elevations of urinary Gb_3. Over both years, a majority of untreated subjects had elevated urinary Gb_3 and tended to have higher values than treated subjects. In conclusion, we show that urinary Gb_3 is elevated in a majority of untreated males and females and trended lower in treated patients. The higher concentrations of urinary Gb_3 observed for males and females during the shortage suggests that ERT dosage impacts urinary clearance of Gb_3. Despite evidence that urinary Gb_3 normalizes early in response to therapy, ~50% of males on ERT had persistent elevations of urinary Gb_3.

POSTER PRESENTATIONS
(Listed Alphabetically by Presenter's Last Name)

CHARACTERIZATION OF SLC6A8 GENE MUTATIONS IN CREATINE TRANSPORTER DEFICIENCY

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SLC6A8 gene loss of function mutations impair the transport of creatine into cells. Based predominantly on mutation data that have been deposited in the Leiden Open Variation Database (LOVD), we conducted a systematic search of studies in the human gene mutation database (HGMD) and PubMed, in which the pathogenicity of SLC6A8 mutation was known. We strongly encourage researchers and clinicians to submit SLC6A8 gene variants to LOVD, as this database is the most expansive published database of pathogenic and non-pathogenic mutations within this gene. We created a database of SLC6A8 mutations, including: location of the mutation, cDNA nucleotide substitution, corresponding protein change, type of mutation and an assigned clinical phenotype. The most frequent type of pathogenic mutations reported were nonsense (43), missense (41) and deletion (32), with 8 distinct *de novo* mutations. It is estimated that approximately 30% of SLC6A8 mutations are *de novo* mutations (van de Kamp et al, 2013). The most commonly reported clinical phenotypes were developmental delay and intellectual disability. No pattern emerged between mutation type or location and decribed phenotype. Mutations appeared to cluster around exons 6 through 12, with exon 9 (16) having the most, next to exon 6 (14), exon 12 (12) and exons 7 and 8 with an equal number of mutations (10 each). These data provide additional insights into this complex disease.

A 14.6 MB DUPLICATION OF XQ25Q27.2 IN A FEMALE: A CASE REPORT

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Previous research has identified a number of males with microduplications within the Xq25q26 region that share common features including short stature, developmental delays and microcephaly. Many of these microduplications have ranged from 0.2 to 4.76 Mb[3]. Lyonization typically mitigates the phenotype for females with microduplications in this area. One additional paper addresses a 1.65 Mb critical region for hemihyperplasia and finger abnormalities[4]. We discuss an African American female who presented to the General Genetics Clinic at the University of Alabama at Birmingham to evaluate for dysmorphic features and failure to thrive at 4 months of age. Growth parameters (height and weight) were at the 50th centile for a one-month-old. Her head circumference has consistently measured below the fifth percentile. Dysmorphic features include posteriorly rotated ears, frontal bossing, retrognathia, and supraorbital hypoplasia. Our patient did not display hemihyperplasia nor finger abnormalities. She underwent a karyotype and array CGH, both of which identified a 14.6 Mb duplication that spans Xq25q27.2. We describe the features seen in our patient as her duplication is significantly larger than others that have previously been documented in other research to further characterize the features that can be associated with a duplication of this size.

POSTER PRESENTATIONS
(Listed Alphabetically by Presenter's Last Name)

USE OF PATIENT REGISTRIES IN A MEDICAL GENETICS CLINIC

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Greenwood Genetic Center[1], University of South Carolina School of Medicine[2], Medical University of South Carolina[3]

Patient registries are often useful to identify individuals with a specific diagnosis so that more can be learned regarding their medical history and to connect them with available research studies. Patients are generally offered the opportunity to enroll based on a broad clinical diagnosis, specific genetic diagnosis, or based on testing history regardless of result (GenomeConnect). Clinicians and researchers typically reach eligible patients through clinic appointments with a subspecialist or through family support groups, while missing those patients who do not present for specialty care or who have a limited online presence. TRANSFORM SC, one of 14 national Institutional Development Awards (IDeA) programs, is building a Pediatric Clinical Research Network in South Carolina, utilizing telehealth technology for the inclusion of rural and underserved communities. As part of this research network, a registry is being developed of patients across the state who may be interested in research participation. Primary care physicians will be provided information to share with their patients, who can enroll regardless of physical location. Targeted medical information, including developmental delay/intellectual disability, autism, birth defects, and known genetic diagnoses, will be collected. Patients who potentially qualify for specific research studies will then be contacted and interest in participation determined. While the registry is developed for all pediatric patients, studies can be targeted to those with conditions that have a specific genetic component. This model provides better access to those patients who may be unaware of current clinical trials or those who have not had the opportunity to be seen by a geneticist.

MINIMIZING FOLLOW-UP TIME FOR CONGENITAL HYPOTHYROIDISM

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Louisiana Office of Public Health, Genetics Diseases Program

Background: Newborn screening tests detect babies with presumptive congenital hypothyroidism (CH) results. Newborns identified with presumptive congenital hypothyroidism require further testing to determine if they are affected. Newborn screening staff are responsible for ensuring that newborns with presumptive results have repeat testing, are evaluated promptly and referred to Pediatric Endocrinology if necessary. The timeliness of follow-up is critical to the diagnosis of newborns. In Louisiana, babies with presumptive congenital hypothyroid results are scheduled for repeat testing. If repeat testing is presumptive for Congenital Hypothyroidism, the newborn is referred to an Endocrinologist. To improve timeliness, a comment was added to the lab report informing physicians to repeat newborn screen and/or perform a serum TSH and FT4 and contact a Pediatric Endocrinologist. **Objective:** Minimize the time from receipt of result to when newborn receives repeat testing, diagnosis or treatment and evaluate the effectiveness of the comment added to the lab report. **Results:** The lab reported 358 presumptive CH babies from June 2015-December 2015 before the comment was added and 341 presumptive CH babies from June 2016 – December 2016 after comment was added. The time from receipt of result to testing, diagnosis or treatment was 15.3 days before comment and 12.7 days after comment was added. A difference of 2.6 days comparing 6 months of data and with more data the number of days could decrease even further. **Conclusion:** Adding a comment for physicians on the newborn screen result may decrease the time between testing and diagnosis for an infant with Congenital Hypothyroidism.

POSTER PRESENTATIONS
(Listed Alphabetically by Presenter's Last Name)

THE EFFECT OF INTACT FOOD SOURCES AND AMINO ACID FORMULA ON RATIOS OF LARGE NEUTRAL AMINO ACIDS IN PATIENTS WITH PHENYLKETONURIA

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Background: Phenylketonuria (PKU) is an autosomal recessive genetic disorder caused by a defect in the liver enzyme phenylalanine hydroxylase (PAH). The lack of PAH activity causes phenylalanine (PHE) to build up in blood and body tissues which is toxic to the central nervous system. Traditional treatment includes lifelong low PHE diet with PHE-free amino acid formula as the primary source of nutrition. It's unknown whether this restricted diet meets U.S. dietary requirements for large neutral amino acids (LNAA), particularly given the need for adequate protein quantity and quality among those with PKU. Also, LNAA are important when treating PKU as they have a common transport system with PHE at the blood-gut and blood-brain barrier. **Objective**: The purpose of this retrospective cohort study is to examine the effect of medical food (specialized PHE-free formula) and intact food protein sources on plasma LNAA in patients with PKU while addressing LNAA dietary requirements. **Methods**: The analysis includes combined participant data from two previous studies conducted at Emory University School of Medicine. Subjects are males and females (N=77) with PKU ages 4-50 years. Variables include demographics, anthropometrics, nutrient intake, genotype/phenotype, blood PHE, and plasma LNAA. A student t-test or Mann Whitney U test will be used to compare LNAA concentration among subgroups (alpha=0.05). Multiple regression analysis will be used to examine the contribution of intact protein to medical food protein ratio and other variables to plasma LNAA. Statistical software is IBM SPSS v. 24. **Results**: To be reported at time of presentation.

PARTICIPATION OUTCOMES IN PILOT LAUNCH OF EMORY GENETICS NUTRITION ACADEMY (eGNA) WEB-BASED CASE CONFERENCE ON ARGINASE 1 DEFICIENCY

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Emory University School of Medicine Department of Human Genetics Metabolic Nutrition Program

Background: eGNA is an online clinical course developed by Emory University to advance clinical practice in the field of inborn errors of metabolism (IEM) with free access to qualified registrants. Pilot launch of EGNA occurred on February 1, 2017, with a one year focus on urea cycle disorder (UCD) cases. **Objective**: To evaluate participation and learning effectiveness to inform future course design and audience applicability. **Methods**: Live case conference was presented by a professional in the field of Arginase 1 Deficiency (ASD1) with accompanying online discussion. Continuing education units (CEUs) were provided to online attendees. Other CEUs could be earned through evidence analysis of a related manuscript and online forum discussions. Pre-test and post-test were provided along with a course evaluation. Additional questions to evaluate audience expertise and promote participation were asked during introduction. Primary data analysis was descriptive, though change between pre- and post-test knowledge scores was analyzed with one-sample t-test (alpha=0.05). **Results**: 135 individuals viewed the registration page, 61 completed course registration, 56 participated in the online case conference, and 42 CEUs were awarded. Majority of attendees were registered dietitians (68%), from medical institutions (75%), and residing in North America (94%). One person completed the journal club evidence analysis. Only 34% of attendees had experience with an ASD1 diagnosed patient. Post-test scores improved from pre-test by 24% ($p > 0.0039$). Overall evaluation was generally positive, although 55% of respondents reported difficulties with registration and site access. **Conclusion:** Findings support community enthusiasm to engage and learn utilizing the Web-based approach for case conferencing. Results will inform our future implementation strategy to improve and simplify registration and site access.

POSTER PRESENTATIONS
(Listed Alphabetically by Presenter's Last Name)

ROLE OF THE METABOLIC DIETITIAN IN MANAGEMENT OF MCADD

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The mortality and morbidity of individuals with medium chain acyl CoA dehydrogenase deficiency (MCADD) has been greatly reduced since the inclusion of the disorder in the recommended uniform newborn screening panel. However. even with early diagnoses, the mortality and morbidity has not been reduced to zero. The goal of management of MCADD is to avoid situations in which the cells must rely solely on stored fats for energy (i.e., fasting). Poor outcomes have been documented in a variety of circumstances where individuals with MCADD were depleted of circulating and stored glucose. While patient families are counseled about fasting in initial visits, it may be that more interactions with healthcare providers, throughout the life cycle, would help remind patients of the circumstances that may lead to fasting and its possible impact on their health. A recent survey of 73 metabolic dietitians revealed that while their clinics saw on average of 2-5 new patients with MCADD annually, only 38% of respondents reported that they were part of the team to see these patients at each clinic visit and to have contact with them between visits. The expertise of metabolic dietitians include: providing appropriate education; maintaining contact and on-going rapport; translating recommendations into daily food intake; assessing nutritional status; anticipating changing needs throughout the life cycle; and advocating for access to appropriate services. These unique skills suggest that the metabolic dietitian should be included as a valuable member of the clinical team working to reduce the mortality and morbidity of MCADD.

MEDICAL MANAGEMENT FOR A SURVIVING INFANT WITH THANATOPHORIC DYSPLASIA

Gooch C, Hurst A
Univeristy of Alabama at Birmingham, Department of Genetics

What medical interventions are appropriate for a patient with a genotype predicted to be life-limiting who continues to live? This case chronicles the ethical and medical complications of a neonate with type 1 Thanatophoric Dysplasia due to *FGFR3* c.742C>T; p.Arg248Cys. At 2 months of age she presented to our hospital for a second opinion for medical management, as an outside hospital had discharged her with a gastrostomy tube, nasal cannula, and home hospice care. At that time her only chronic problem was a 1-liter nasal cannula oxygen demand, aggravated by a recent Rhinovirus infection. The child was otherwise well-appearing, alert, smiling, and interactive. A brain MRI showed cervical stenosis, a known complication of skeletal dysplasias. Discussions with the mother and neurosurgery, neonatology, palliative care, and genetics were challenging due to few case reports of prognostication for survivors with this condition, and national experts were contacted for guidance. Due to her lack of other life-limiting complications, cervical decompression was felt to be justified as in patients with achondroplasia. After a multidisciplinary meeting, she was taken for successful decompression and was discharged. However, at 4 months of age she represented with possible seizure activity and was found to have foramen magnum compression and associated respiratory distress requiring mechanical ventilation. At that time family declined further intervention and requested withdrawal of ventilator support.

POSTER PRESENTATIONS
(Listed Alphabetically by Presenter's Last Name)

BENEFICIAL EFFECTS OF CARVEDILOL WITH ENZYME REPLACEMENT THERAPY IN POMPE DISEASE

Han S[1], Haynes AC[2], Li S[1], Kishnani PS[1], Steet R[2], and Koeberl DD[1]*
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Pompe disease (glycogen storage disease type II) is caused by the deficiency of lysosomal acid α-glucosidase (GAA) leading to progressive myopathy. Enzyme replacement therapy (ERT) with recombinant human (rh) GAA improved outcomes, although limitations remain including inefficient uptake of rhGAA in skeletal muscle linked to low expression of cation-independent mannose-6-phosphate receptor (CI-MPR). Agents that cause muscle hypertrophy by increasing insulin-like growth factor 1 expression, such as the □2-agonists clenbuterol and albuterol, can increase CI-MPR-mediated uptake of rhGAA with therapeutic effects in skeletal muscle. We hypothesized that alternate agents with hypertrophic effects might be efficacious in combination with ERT. Four such agents were evaluated in mice with Pompe disease (oxandrolone, losartan, growth hormone, and carvedilol). Carvedilol, a □-blocker, increased muscle strength ($p<0.05$) but reduced GAA activity following ERT ($p<0.01$ in diaphragm, $p<0.1$ in quadriceps). Intriguingly, combining carvedilol with albuterol treatment enhanced the biochemical correction from ERT ($p<0.05$ in diaphragm) and resulted in a two-fold increase in CI-MPR levels in skeletal muscle. Uptake experiments using the same drugs in Pompe fibroblasts also confirmed the synergistic effect. Administration of drugs alone had minimal effect, with the exception of losartan that increased glycogen storage ($p<0.1$ in quadriceps) and mortality either by itself ($p<0.05$) or in combination with ERT ($p<0.1$). In summary, we demonstrate that the □-blocker carvedilol had beneficial effects when used in combination with albuterol during ERT and should be considered as a β-blocker for Pompe patients.

A SYSTEMATIC APPROACH TO THE DIAGNOSIS OF PEDIATRIC PATIENTS WHO PRESENT WITH METABOLIC CRISIS IN THE EMERGENCY DEPARTMENT

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Inborn errors of metabolism (IEMs) affect approximately 1 in 500 newborns. Some of these patients will present emergently to the hospital in metabolic crisis. Having a systematic approach to rapidly obtaining a diagnosis for these patients is critical to improving outcomes in the emergency department when treating metabolic crisis. Vomiting or fasting related to infection are a common trigger of metabolic decompensation in a child with disorders of protein and carbohydrate metabolism or energy production. Sudden increases in protein consumption or catabolism of body protein stores can trigger a metabolic crisis in children with urea cycle disorders, organic acidurias, aminoacidopathies, and protein metabolism disorders. Increased dietary fat consumption will trigger symptoms in children with fatty acid oxidation disorders, lipoprotein lipase deficiency, and glycerol intolerance. Episodes of metabolic crisis can occur in children with galactosemia when they consume milk products. Fructose intolerance will cause symptoms in children unable to metabolize fructose. Exercise can also induce metabolic decompensation in children with a variety of disorders that result in an insufficient energy supply to muscle cell such as disorders of fatty acid oxidation or muscle glycogenolysis. Additionally, medications can trigger a crisis in children with any of the porphyrias or Glc-6-P-dehydrogenase (G6PD) deficiency. Being alert to these common triggers may help to narrow the focus rapidly during a metabolic emergency. The purpose of this abstract is to summarize a practical approach to narrowing the differential diagnosis in the emergency department when a pediatric patient presents to the emergency department in metabolic crisis.

POSTER PRESENTATIONS
(Listed Alphabetically by Presenter's Last Name)

MABRY SYNDROME: USING A COMMON BIOMARKER, ALKALINE PHOSPHATASE, TO IDENTIFY RARE NEUROMETABOLIC DISEASE

Logan R[1], Foley A[2], Shankar P[3], and Li H[2]
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Mabry syndrome (also known as Hyperphosphatasia with Mental Retardation Syndrome, HPMRS) is a rare neurometabolic condition that manifests predominantly with neurological symptoms, including intellectual disability, developmental delay, seizures, and constitutively high serum alkaline phosphatase (ALP). ALP is commonly ordered in the hospital setting as part of a comprehensive metabolic profile. Elevation of ALP is often interpreted as a non-specific biomarker of bone, liver or GI disease. Due to lack of provider knowledge about ALP as a biomarker, combined with the heterogeneous nature of this rare genetic condition, persons with Mabry syndrome have the potential to go undiagnosed. There are only about 55 reported cases in the literature. The purpose of this study is to use a retrospective chart review approach to understand how common Mabry syndrome is in the Children's Healthcare of Atlanta (CHOA) patient population and to improve medical care by identifying undiagnosed persons. Utilizing this approach, 2823 children with ALP above 500 IU/L from 2012-2016 were identified. After exclusion of individuals with other explanations for high ALP level and other clinical findings, 12 children highly suspect for Mabry syndrome were ultimately identified. Referral to genetics clinic for evaluation is done in collaboration with the primary care physician. A diagnosis of Mabry syndrome can be beneficial to both clinician researchers and patients as it provides a unifying diagnosis for the patient and an opportunity to learn more about this heterogeneous condition. One person has been evaluated as a part of this study and recruitment is ongoing.

A STUDY OF THE GLYCEMIC PROFILE IN GLYCOGEN STORAGE DISEASE USING CONTINUOUS GLUCOSE MONITORING

Pendyal S, Herbert M, Rairikar M, Austin S, Benjamin R, Kishnani P
Duke University Medical Center, Durham, NC

The management of many glycogen storage diseases (GSD) involves adherence to a strict diet and checking finger stick blood glucose (BG) levels multiple times daily to maintain euglycemia. To study the metabolic control and variations in glycemic profile, continuous glucose monitoring (CGM) using the DexCom G4 real time device was performed in 17 GSD patients (5 with GSD Ia, 2 with GSD Ib, 6 with GSD IIIa, and 4 with GSD IX, ages 5 to 55 years). Patients were monitored for an average of 8.8 days. CGM data was available for a total of 142 days with 33,162 glucose readings (average 288 readings/ patient/ day). Patients spent an average of 90% (range 88-94%) time in the target glucose range of 70-150 mg/dl. GSDI patients spent less time with euglycemia (88%) when compared to GSD III (98%) and GSD IX (89%) and more time in hypoglycemia, with glucose levels <70 mg/dl (9%), <60 mg/dl (3%) and <50 mg/dl (1%) compared to those with GSD III and IX. The most vulnerable times for hypoglycemia were early morning (2-6 am), mid-day and at night (9-11 pm) across all GSD types. Interestingly, hyperglycemia was also seen in all GSD types with higher incidence (15%) in GSD IX than GSD III (7%) and GSD I (8%), possibly reflecting overtreatment with diet or corn starch. CGM with concurrent dietary adjustments improved metabolic parameters and stabilized glucose levels in three patients. Studying the glycemic profile in GSD with CGM can provide valuable information for optimizing treatment.

POSTER PRESENTATIONS
(Listed Alphabetically by Presenter's Last Name)

MEDICAL NUTRITION THERAPY FOR PREVENTION FOR INHERITED METABOLIC DISORDERS

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The Medical Nutrition Therapy for Prevention Program (MNT4P) was established through the Emory Genetics Metabolic Nutrition Program and the Georgia Department of Public Health to bridge coverage gaps by providing consistent medical nutrition therapy for patients with inherited metabolic disorders (IMD) in Georgia. MNT4P aims to create a national model for healthcare delivery and longitudinal data collection for IMD. In Year 1, the objective for MNT4P was to build a patient-centered programmatic framework through strategic planning and stakeholder collaboration. In order to build an effective programmatic framework, multiple public health methodologies were utilized. These key strategies included evaluation, interviewing, coalition building, advocacy and systems development. An informal evaluation of the metabolic community was conducted to understand barriers to care and how they are addressed in other states. Metabolic dietitians at the Emory Clinic similarly incorporated basic interview questions into their nutrition assessment to gauge patient needs in Georgia. The Georgia Medical Foods Advisory Board was then developed as a coalition of stakeholders in the metabolic community charged with strategizing best practices for program development. In addition to the Advisory Board, the MNT4P team has actively collaborated with consultants on advocacy and systems development, with the intention of designing an information system accessible by both patient and provider/professional. This presentation will include a summary of the programmatic framework and gains made in Year 1. MNT4P will be the first program to both serve patients and integrate insurance, medical foods, and health outcomes data to drive quality improvement and inform policy development.

IDENTIFICATION OF EDUCATIONAL GAPS IN GENETIC KNOWLEDGE VIA AN INNOVATIVE AND INTERACTIVE WEBSITE

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Purpose: To evaluate existing genetic knowledge gaps in individuals living with a genetic condition and learn how they interact with an informational website on topics such as natural history, disease management, and treatment options related to genetic conditions. **Methods**: After providing consent, participants completed a brief survey containing demographic information and knowledge questions. Participants were then asked to use the interactive ThinkGenetic patient educational site and allow us to anonymously track which areas of the website they visited and the questions asked. **Results**: A total of 74 participants completed the demographics and knowledge survey. Website users most often sought practical information about living with genetic conditions and where they can find more information about the condition in question. Individuals living with a genetic condition were not significantly more likely to know how the genetic condition is inherited than those who are a family member of someone living with a genetic condition. **Conclusion**: Commonly asked questions suggest that providers need to focus on, provide additional resources for, or emphasize specific topics including: practical information about living with genetic conditions and where they can go for additional information and resources.

POSTER PRESENTATIONS
(Listed Alphabetically by Presenter's Last Name)

IMPROVED METHOD FOR DIAGNOSIS OF CEREBRAL CREATINE DEFICIENCY SYNDROMES IN BIOLOGICAL MATRICES BY ULTRA-PERFORMANCE LIQUID CHROMATOGRAPHY ELECTROSPRAY IONIZATION TANDEM MASS SPECTROMETRY

Stiles AR[1,2], Seifts A[2], Kilibarda N[3], Goldstein JL[4], Koeberl DD[1,2], and Young SP[1,2]
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Cerebral creatine deficiency syndromes (CCDS) comprise a group of treatable inborn errors of creatine biosynthesis: arginine:glycine amidinotransferase (*AGAT*) and guanidinoacetate methyltransferase (*GAMT*) deficiencies, and a functional defect in the creatine transport: creatine transporter deficiency (*SLC6A8*). CCDS have overlapping clinical features which include intellectual disability, speech delay, seizures, abnormal movements, and autistic-like behavior. Neurological involvement can be prevented with early identification and therapeutic intervention. Biochemical testing for CCDS is routinely performed by analysis of creatine and guanidinoacetate in plasma and/or urine for diagnosis of AGAT and GAMT deficiencies whereas urinary creatine to creatinine ratio is utilized for the successful diagnosis of creatine transporter deficiency. We describe an improved high-throughput method for simultaneous analysis of creatine, guanidinoacetate (GAA), and creatinine from human plasma and urine without derivatization by stable isotope dilution ultra-performance liquid chromatography tandem mass spectrometry (UPLC-MS/MS). Less than 12% difference was observed for all analytes in both urine and plasma matrices after reanalysis of samples with the modified method. Furthermore, we describe the utility of dried urine spots (DUS) as an alternative to liquid urine, which upon normalization to creatinine, DUS GAA and DUS creatine concentrations were in good agreement (within ± 12.5%) with measured values in liquid urine. Finally, we present the clinical utility of this method in the diagnosis of two male siblings with a positive family history of x-linked creatine transporter deficiency and show elevated urinary creatine at <24 hours (1225 mmol/mol creatinine; nl: 3.8-947) and day of life 6 (2485 mmol/mol creatinine; nl: 3.8-947), respectively.

HOMICIDAL TENDENCIES AT EARLY PEDIATRIC AGE IN MALES CARRYING VARIANTS IN IQSEC2

Walano N, Preston G, Dvorak C, Nelson S, Andersson H, Morava E
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IQSEC2, a gene that has been implicated in non-syndromic X-linked intellectual disability, has also recently been associated with behavioral abnormalities such as autistic behavior, self-injurious behavior, and repetitive stereotypic movements. Mutations in this gene are mostly de novo. Many carrier females are unaffected, although, some have been found with mild to moderate intellectual disability. Next generation sequencing in a group of individuals with suspected seizures and neurodevelopmental disorder revealed variants of unknown significance in three cases in the IQSEC2 gene. All of these variants were well-conserved missense amino acid alterations predicted to impact secondary protein structure. We evaluated the molecular segregation of the reported variants and clinical features in these three families. We confirmed the variant in the mother and sister of our index case and in the mother of the second and third case as well. All of the individuals carrying the IQSEC2 variants have overlapping psychiatric features. The male individuals have shown aggression and violent tendencies, while the female individuals had milder presentation in the form of depression and suicidal behavior. Additionally, in two of the three male individuals we found large, broad big toes and a bulbous nose as observed in previous individuals with proven IQSEC2 pathogenic variants; these mild dysmorphisms have not been evaluated in the third individual and were not noted in the carrier females. Individuals with pathogenic variants in the IQSEC2 gene range in severity in their clinical presentation; from highly intellectually disabled and resembling Rett Syndrome (microcephaly, language regression and hand stereotypies) to seizures with mild developmental delay. Here we describe profound behavioral abnormalities with significant aggression, suspected seizures, autism in carrier males, and psychopathology in carrier females. We suggest that these symptoms are associated with IQSEC2 related disease.

POSTER PRESENTATIONS
(Listed Alphabetically by Presenter's Last Name)

A RETROSPECTIVE CASE PILOT PROJECT BETWEEN FDNA AND THE GREENWOOD GENETIC CENTER

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Greenwood Genetic Center

FDNA uses facial analysis, deep learning, and artificial intelligence to transform big data into actionable genomic intelligence to improve diagnostics and therapeutics. FDNA's software, Face2Gene, facilitates comprehensive and precise genetic evaluations through the capture and analysis of facial data and characteristic phenotypes relevant to rare diseases. The Greenwood Genetic Center (GGC), founded in 1974, is a nonprofit organization that provides care for families impacted by genetic disease. The home campus is located in Greenwood, SC, and provides clinical genetic services, diagnostic laboratory testing, educational programs, and medical genetics research. Additionally, four satellite offices extend clinical services across the state. GGC has routinely stored data electronically since 2000. Prior to this, large amounts of data were stored in paper charts, printed photographs, and 35mm slides. This non-digitalized data is accessible, though much of it remains in storage. In the current electronic data storing system, information is stored on approximately 80,000 patients seen clinically. In this same system, photographs and laboratory data is also stored for a large percentage of patients seen clinically. FDNA and GGC have initiated a pilot project aiming to create a clinical data warehouse that is able to electronically store clinical documentation, phenotype data, photographs, laboratory results, and diagnosis. Through this process, data in a non-digital format will be digitalized and stored, improving accessibility and utility for case management and downstream research and analysis. Face2Gene's learning database will be able to extract non-identifiable data, including facial and phenotypic information, to train and enhance the Face2Gene software, benefiting the genetics community worldwide.

URINARY FREE OLIGOSACCHARIDE ANALYSIS AS A POTENTIAL BIOMARKER FOR MUCOLIPIDOSIS TYPES II, II/III, IIIA/B

Wood T, Huang R, Cathey S, Pollard L
Greenwood Genetic Center

Mucolipidosis (ML) types II, II/III, and IIIα/β are allelic lysosomal storage disorders caused by mutations in the *GNPTAB* gene that result in a deficiency of GlcNAc phosphotransferase. ML II patients have a severe clinical course presenting before a year of age with bone abnormalities, intellectual disability and growth retardation. ML II/III patients have a more intermediate presentation while ML IIIα/β patients can present later in childhood with only bone involvement. Measurement of urinary free oligosaccharides (FOS) is one clinical test which can be used to identify ML patients. Our laboratory has developed a novel UPLC MSMS assay for seven FOS species which shows 100% sensitivity for the detection of associated glycoproteinoses. As FOS analysis has been previously demonstrated to be abnormal in ML patients, we measured these seven FOS in 75 urine samples from a cohort of ML patients. Three of the seven FOS species show significant elevations in ML patients as compared to controls suggesting they, collectively, can be used as a biomarker for this disease. We then stratified our patient cohort based on clinical classification of ML II, ML II/III, ML IIIα/β. A statistically significant difference in the levels of these three FOS species was found between patients with ML II and ML II/III versus patients with ML IIIα/β. Because these FOS levels correlate with clinical presentation of ML, we hypothesize these species and/or additional FOS may serve as good biomarkers to evaluate the efficacy of future therapeutics for ML.

POSTER PRESENTATIONS
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ARTICULAR CARTILAGE GLYCOSAMINOGLYCAN CONCENTRATIONS IN A DOG MODEL OF MPS TYPE I

Zhang H[1], Ellinwood NM[2], Snella EM[2], Kan S-H[3], Dickson PI[3], Wang R[4], and Young SP[1]
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[2]Department of Animal Science, Iowa State University, Ames, IA;
[3]Los Angeles Biomedical Research Institute at Harbor-UCLA Medical Center, Torrance, CA;
[4]Division of Metabolic Disorders, CHOC Children's Hospital, Orange CA &, Department of Pediatrics, University of California Irvine School of Medicine, Irvine, CA.

Objective: To evaluate articular glycosaminoglycan concentrations in a canine model of mucopolysaccharidosis type I (MPS I). Background: MPS I is a lysosomal storage disorder caused by □-iduronidase deficiency, resulting in glycosaminoglycan (GAG) accumulation. Approved therapies aimed at restoring α-iduronidase activity, including enzyme replacement therapy (ERT) or hematopoietic stem cell transplantation, inefficiently target joints. The failure to reduce GAGs in cartilage and bone results in progressive orthopedic complications. A canine model of MPS I has been used to investigate the efficacy of ERT applied via intravenous, intra-thecal and intra-articular routes. We sought to characterize chondroitin sulfate (CS), dermatan sulfate (DS) and heparan sulfate (HS) concentrations in articular cartilage in this model using methanolysis combined with ultraperformance liquid chromatography-tandem mass spectrometry. Methods: Articular cartilage samples were obtained from bilateral shoulder, hip, and stifle joints of MPS I (n = 3) and control (n = 3) animals. Cartilage was protein digested and CS, DS, and HS were analyzed by a published method. Results: MPS I dogs had significantly higher DS and HS concentrations ($p<0.05$) in all sites (shoulders, hips, stifles), and significantly lower CS concentrations in all sites except the stifles. A difference was also observed between the different joints within a group (MPS I or controls), wherein GAG concentrations were highest in shoulders and lowest in stifles. Conclusions: This work further characterizes articular cartilage changes in a dog model of MPS I, which will better inform the impact of new treatment strategies aimed at correcting α-iduronidase deficiency within joints.

NOTES

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www.progenygenetics.com

Progeny Clinical saves you time with patient-entered family history questionnaires, integrated risk models, custom letter generation and streamlined genetic testing. Coming soon is the ability to generate custom letters and dictations.

Retrophin
3721 Valley Centre Drive
San Diego, CA 92130
Phone: 760-260-8600
Representative: Shawn Hayes
www.retrophin.com

Retrophin is a biopharmaceutical company dedicated to delivering life-changing therapies to people living with rare diseases who have few, if any, treatment options.

SERN at Emory University
2165 North Decatur Road
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Phone: 404-778-8521
Representative: Rosalynn Blair
www.southeastgenetics.org

Advertising and educational information about Emory SERN's workgroup projects.

Thermo Fisher Scientific
3450 Central Expressway
Santa Clara, CA 95051
Phone: 408-731-5122
Representative: Luke Dirscherl
www.thermofisher.com

Thermo Fisher offers the FDA-cleared CytoScan® Dx Assay, first-of-its-kind test to help diagnose developmental delays and intellectual disabilities in children from postnatal blood samples. Visit our booth to learn more about this breakthrough product.

Vitaflo USA
211 North Union St., Suite 100
Alexandria, VA 22314
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