



## JULY - SEPTEMBER 2023 NEWSLETTER

# SERN/SERGG 2023 ANNUAL MEETING

The SERN/SERGG joint Annual Meeting took place July 13-15, 2023, in Charleston, SC. Here are some of the highlights from the SERN portion of the meeting.



Dr. Joseph Muenzer speaking at the Paul M. Fernhoff Memorial Lecture.

## TELEMEDICINE WORKGROUP

### What You Need to Know About the Digital Divide - Amy Sheon, PhD, MPH.

In addition to access difficulties in rural areas, Dr. Sheon discussed persisting racial disparities in internet access in inner city areas. She described the 2021 Infrastructure Investment and Jobs Act, which included \$65 billion investment in broadband upgrades. Part of this money is allocated to the Affordable Connectivity Program ([affordableconnectivity.gov](https://affordableconnectivity.gov)) which provides \$30 per month toward internet service for households meeting certain financial criteria, or for households with a member who participates in certain government assistance programs (such as SNAP, Medicaid, WIC, etc). Dr. Sheon also presented at the SERN Family Alliance Breakout Session, where the discussion included the healthcare record-related issues of privacy, access, and ease of use.

### Patient Perspectives on Telemedicine - moderated by Rossana Sanchez, MD.

Caregivers of children with genetic conditions participated in a panel discussion. Benefits of telemedicine discussed include less loss of work hours, comfort of the child in their own home setting, and reduced travel burden on the family. Telemedicine can be facilitated for families with detailed instructions on how to download and utilize the appropriate app.

## PLATFORM SESSION 1

### DNA Methylation Episignatures: Diagnostic Biomarkers for Rare Diseases and Beyond - Bekim Sadikovic, PhD.

The presenter discussed epigenetics definitions and mechanisms, and then episignatures. The uses of episignatures discussed included methylation pathogenicity scores, new syndrome identification, identification of genes involved in microdeletion/duplication syndromes, understanding penetrance, and classifying patients with uncertain diagnoses. The clinically-validated genome-wide methylation testing, EpiSign, is used for the reclassification of variants of uncertain significance. Current Health System Studies underway were discussed.

### Mendelian Disorders of the Epigenetic Machinery: Clinical Insights and New Discoveries - Jill Fahrner, MD.

Dr. Fahrner discussed chromatinopathies, or mendelian disorders of the epigenetic machinery (MDEMs). Categories of MDEMs include writers, erasers, readers, and remodelers. Most MDEMs are autosomal dominant, and are often due to de novo mutations or haploinsufficiency. Phenotypes vary but often include intellectual disability and growth abnormalities. The Epigenetics and Chromatin Clinic at Johns Hopkins has identified 115 patients with MDEMs in the past five years, due to 26 distinct genes.

### Dr. Paul M. Fernhoff Memorial Lecture.

In honor of Dr. Paul M. Fernhoff, SERN hosted a memorial lecture focused on newborn screening and related disorders. The lecture was given by Dr. Joseph Muenzer who presented on the impact of new treatment strategies and newborn screening on Hunter syndrome (MPS II). The lecture included an overview of MPS disorders and their pathophysiology. A specific focus was on Hunter syndrome, an ultra-rare X-linked recessive disorder, with discussion of molecular genetics, current treatment options (hematopoietic stem cell transplantation or IV enzyme replacement therapy). Novel treatment strategies that are under investigation include medications that enhance blood-brain barrier penetration, gene therapy, and intrathecal enzyme replacement therapy. MPS II was added to the Recommended Uniform Screening Panel (RUSP) in 2020.

# SERN/SERGG 2023 ANNUAL MEETING HIGHLIGHTS CONT'D

## SERN FAMILY ALLIANCE BREAKOUT SESSION

### Supports that Family Leaders Need to Make Them Successful Partners - Molly Martzke.

Molly delivered a lecture discussing the influential role of families in advocating for themselves and others. She skillfully recounted her own journey in family leadership and advocacy, from her involvement with Wisconsin State Advisory Committee in implementing newborn hearing screening to becoming a family leader within the Wisconsin Children and Youth with Special Health Care needs (CYSHCN). She underscored the significance of active participation, regardless of the scale, in addressing family needs through advocacy. Her session inspired all attendees to share their personal experiences and engage in a fruitful exchange of ideas.

### Genetic Testing for Families - Gwen Gunn, PhD, MS, CGC.

Dr. Gunn provided a high-level overview of genetic testing, identifying the key components of personal genetic test results, and discussing the why, who, how and what of genetic testing and personalized medicine. She facilitated an interactive session with family members, addressing any inquiries related to genetic testing and its potential consequences.

## TASTE OF eGNA

### Taste of eGNA: Long Chain Fatty Acid Oxidation Disorders and the Role of MCT/Triheptanoin

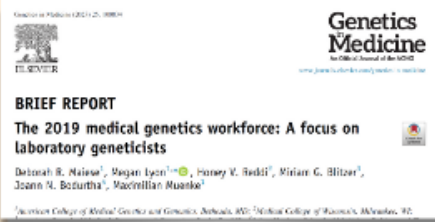
The eGNA team hosted their first hybrid educational dinner demonstrating the Genetic Nutrition ECHO educational program, an instance of Project ECHO. Rosalynn Blair, MA kicked off the session. Lindsay Ryan, MS, RD, LD shared information about the history and current state of the Electronic Genetic Nutrition Academy (eGNA) and the subset of eGNA that was the main focus of the event, eGNA's Genetic Nutrition ECHO. To provide background knowledge before the case, Rani Singh, PhD, RD, LD shared a presentation on the nutritional management of patients with long chain fatty acid oxidation disorder (LCFAOD). Lastly, Stephanie Hacker, MS, RD/N, LD/N, CSSD presented a case of a LCFAOD patient and reviewed the patient's history and treatment with triheptanoin. Surekha Pendayal, Msc, MEd, RD, FAND joined the hub team as an expert panelist and Hosana Nagasaka, MPH, RD wrote down the case recommendations to send to the case presenter afterwards. As part of the demonstration, both the in-person and Zoom audiences were involved in asking clarifying questions and providing recommendations on the case.

## New Publication from the National Coordinating Center for Regional Genetics



The 2019 medical genetics workforce: A  
focus on laboratory geneticists

The National Coordinating Center for the Regional Genetics Networks (NCC) is pleased to announce the publication of "The 2019 medical genetics workforce: A focus on laboratory geneticists". The paper highlights the demographics of the laboratory geneticist workforce from our 2019 workforce survey. Learn more by reading the article at:  
<https://bit.ly/2019LabGeneticist>



# MEET SOME OF OUR REGIONAL GENE-ies!

FAMIL TO FAMILY NETWORK PROGRAM DIRECTORS

## FLORIDA



## LISA MATH

**Family S.T.A.R. Program - Family Network on Disabilities** - At Family Network on Disabilities, we strive to provide families and self advocates with support, training and education, and resource linkage so they can advocate effectively for their loved ones. We are always reviewing, creating, and curating resources and materials to share with families. We serve as a conduit of information in the education, medical, and mental health systems in our state. We also work on the state and national level to support organizations, and to collaborate with federal and state agencies to ensure the family voice is heard in program development and review.

## SUSAN BROWN

**Parent to Parent of Georgia (P2PGA)** - At P2PGA we provide families with one-on-one assistance and offer free trainings on various healthcare, education, and transition-related topics. We believe these are some of the biggest challenges in our state: 1. Lack of Medicaid expansion, 2. Severe lack of trained childcare providers, specifically serving children with special healthcare needs -- several years ago two medically fragile daycare providers shut their doors, forcing many families to make hard choices regarding working and putting single parents in an untenable situation, and 3. the significant lack of adequate pediatric healthcare providers in Georgia.

## GEORGIA



## LOUISIANA



## JULIA FOLSE

**Louisiana Family to Family Health Information Center** - At our state's resource center, our primary focus is on assisting families in navigating systems and accessing community services easily. We dedicate significant efforts to help families understand and navigate complex systems such as healthcare, education, social services, and disability-related programs. Our team is well-versed in the intricacies of these systems and can guide families through the processes, requirements, and available options. We offer personalized assistance to address specific needs and challenges, ensuring that families have the information and tools to advocate for their loved ones effectively.

# MEET SOME OF OUR REGIONAL GENE-ies!

FAMIL TO FAMILY NETWORK PROGRAM DIRECTORS

## NORTH CAROLINA



### CHRISTY MOORE

**Exceptional Children's Assistance Center (ECAC)** - The work that the Family to Family Health Information Center grant allows for has been a passion area of mine since my youngest daughter was born in 2010. I had concerns very early on about her development, and unfortunately was given the “wait and see” advice by her pediatrician. When my daughter was around 3 years old, I became passionate about sharing information with other families so they could make timely, informed decisions about their child’s care. We were awarded a grant in June 2022 and we have been working to support families and providers in NC by providing one-on-one support, in-person training and webinars, fact sheets, and facilitating community partnerships.

## AMY HOLBERT

**Family Connection of South Carolina** - As a pediatric social worker, I made referrals to Family Connection’s programs, trainings and abundant informational materials however, I never imagined that I would become one of the parents that would be on the receiving end of Family Connection’s parent support program. My youngest son was preterm and had an extended stay in the NICU. After a particularly rough night for my son, the Family Support Specialist from Family Connection found me alone and crying in the lobby. She wasn't afraid to be there with me in my fear, and I'll never forget what she did for me that day. This is what makes Family Connection of SC different from other support and educational programs, we are staffed by highly trained, skilled parents of children with a disability, chronic health condition, or behavioral health diagnosis.

## SOUTH CAROLINA



## TENNESSEE



### CAROL WESTLAKE

**Tennessee Disability Coalition** — Families face many challenges as they work to navigate the systems and support they need to assure that their children have the best opportunities to grow up healthy, happy, and as independent as possible. With all of the complexities and barriers they face, getting access to reliable, useful, timely, and accurate information is a one big challenge that the F2Fs work to address. In addition to helping families learn to successfully navigate health systems, and actively engage with health care professionals, our focus is on elevating the family voice in all aspects of health care. We work to improve the responsiveness of health care systems and improve the quality of care for Children and Youth with Special Health Care Needs.

# SERN-FUNDED RESEARCH

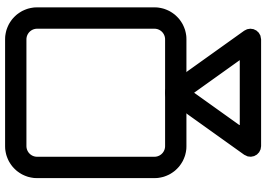
In 2015, SERN and Genetic Metabolic Dietitians International (GMDI) published evidence- and consensus-based guidelines for the nutrition management of phenylketonuria (PKU). In 2018, the US Food and Drug Administration approved the use of pegvaliase (Palynziq® BioMarin, Novato, CA) as an alternate pharmacotherapy for management of PKU. Therefore, SERN and GMDI convened a workgroup to formally assess the research literature and current clinical practices to update their PKU nutrition management guidelines to incorporate recommendations related to pegvaliase. The resultant guidelines have recently been published in *Orphanet Journal of Rare Diseases*, and they include (1) initiating a pegvaliase response trial, (2) monitoring therapy response and nutritional status, (3) managing pegvaliase treatment after response to therapy, (4) education and support for optimal nutrition with pegvaliase therapy, and (5) pegvaliase therapy during pregnancy, lactation, and adolescence. The guidelines are also available online ([managementguidelines.net](http://managementguidelines.net)) along with a Consumer Summary in English and Spanish and a practitioner toolkit.

Cunningham A, et al (2023) [Nutrition management of PKU with pegvaliase therapy: update of the web-based PKU nutrition management guideline recommendations](#). *Orphanet Journal of Rare Diseases* 18(1):155. doi: 10.1186/s13023-023-02751-0.

# 2023 Metabolic Camp Report



This year, the 29th Annual Metabolic Camp was held June 19-24. This camp was the first in-person camp to be held in the last three years, and both campers and counselors were happy about it. There were 30 campers who travelled from all over the country to attend the camp. The campers were of all different ages, most being women and girls with PKU, and some with MSUD. Campers and counselors alike participated in team-building activities and informational classes to educate campers on nutritional management and reproductive health. Some highlights of the week included a cooking class, a belly-dancing class, and a maternal health panel. Campers also had the opportunity to go on fun excursions, such as the Museum of Illusions in Atlantic Station. Overall, both campers and staff were grateful for the opportunity to connect and have fun with one another in person again. We are happy that this year was a success and excited to welcome back returning campers and meet new campers at next year's camp!



## COMING SOON:

Videos from recorded presentations from the SERN/SERGG 2023 Annual Meeting will be available online soon.

## SAVE THE DATE!

### SERN/SERGG ANNUAL MEETING

July 11-13, 2024

Renaissance Marriott Hotel

Asheville, North Carolina

## Impact of 2022 Formula Shortage

In February 2022, there was an acute powdered formula shortage, due to the six-month halt in production of Abbott Nutrition's main manufacturing facility. Although this impacted almost all infants across the U.S., it also had a substantial impact on those with inherited metabolic disorders (IMD), who utilize metabolic formulas for their dietary therapy. Medical Nutrition 4 Therapy (MNT4P) found that 40% of their patients managed with medical food in Georgia utilized Abbott products prior to the shortage. Due to having to adopt new medical foods, these patients experienced dietary intolerance and gastrointestinal issues – this further resulted in decreases in treatment adherence and increase in hospitalizations. In response to these crises, different organizations, including MNT4P and the Southeast Regional Genetics Network (SERN), collaborated with key stakeholders in medical food production and dispersal, to mitigate the effects of the shortage. For future prevention, it is suggested to diversify formula consumption from a young age, and for the Genetics Metabolic Dietitians International (GMDI) to create a committee that addresses emergencies in the IMD community, to better prepare for any future shortages.

# LOW PROTEIN RECIPE

## MASON JAR – MANGO SPRING ROLL

### INGREDIENTS

#### Spring Roll:

- 1/2 bundle vermicelli (mung bean or green bean) low-protein noodles, cooked
- 1/4 cup carrots, raw, chopped
- 1/4 cup sweet red pepper, raw, chopped
- 1/2 cup mango, diced
- 10 small leaves mint, raw, chopped
- 3 large leaves basil, raw, chopped

#### Honey Soy Lime Sauce:

- 2 tsp lime juice, fresh
- 1 tsp reduced sodium soy sauce
- 1 tsp water
- 1 tbsp honey

### INSTRUCTIONS

- After cleaning the food preparation area you will be using, gather the equipment and ingredients you need for this recipe.
- Wash your hands with soap and water.
- For the vermicelli noodles, follow the package instructions. Measure the weight of the cooked noodles and divide in half. You only need ½ the noodles for this recipe. Set aside.
  - a. *For our preparation, boiling water is poured over the dry noodles and the noodles soaked for 6 minutes. Noodles are then drained and rinsed with cold water. With clean scissors, the noodles are cut so they were in bite-size pieces.*
- Once you have measured and chopped the ingredients, you are ready to make the Honey Soy Lime Sauce for your Mango Spring Roll.
- To a bowl, add the lime juice, reduced sodium soy sauce, water and honey. With the whisk, mix well and set aside.
- To the mason jar, add 2 TBSP of the Honey Soy Lime Sauce.
- Add your vegetables (red cabbage, carrots, sweet red peppers), followed by the mango, mint and basil.
- The cooked vermicelli noodles will be the last item to be added to the jar.
- Cover and shake well to make sure the sauce is coating the noodles, vegetables, mango, basil and mint. Enjoy!

### ADDITIONAL INFORMATION

#### Meal Planning Tip

You can make 2 Mango Spring Roll servings since you are preparing a bundle of the vermicelli noodles, but only half is used for the recipe. If not consuming the mango spring roll immediately, have the Honey Soy Lime Sauce on the side to be added once ready to eat.

#### Nutrition Facts

Yield: 1 mason jar (16 oz)

Nutrition Facts (Per 1 Serving):

PHE: 89 mg  
LEU: 129 mg  
Protein: 2.1 g  
Carbohydrates: 56 g  
Calories: 220

Recipe created by Kristen Narlow, MS, RDN, LDN, CC. Nutritional analysis conducted by RDs at Emory University. Nutritional analysis based on the products used at the time of analysis. Subject to change.

#### Resources

[Safe Recipe Guide](#)  
[CDC Fruit & Vegetable Safety](#)  
[How to prepare mango](#)

## UPCOMING EVENTS

- September 8-10: [Georgia PKU Connect Family Camp](#). Adults with PKU and families with children that have PKU ages 4-18 are invited for a weekend of low protein cooking, outdoor activities such as bike riding, fishing, and horseback riding. Anyone interested should email [camp@georgiapku.org](mailto:camp@georgiapku.org) by Friday, August 25th.
- eGNA: Journal Club on Oct. 27 at 12-1pm EDT and Case Conference on Nov. 3 at 12-1pm EDT

## AWARENESS CALENDAR

### August

- **Spinal Muscular Atrophy (SMA) Awareness Month**

### September

- **National Sickle Cell Month** with the [Sickle Cell Disease Association of America](#)
- **Newborn Screening Awareness Month** with the [Health Resources and Services Administration](#)
- **Tay-Sachs Awareness Month**
- **September 18-24: World Mitochondrial Disease Week**



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