SERN SOUTHEAST REGIONAL GENETICS NETWORK

APRIL - JUNE 2023 NEWSLETTER

HIGHLIGHTS FROM the MCADD Summit

On Saturday, February 25, SERN co-hosted the MCADD Patient & Family Education Summit with Providers. The summit was also supported by the Medical Nutrition Therapy for Prevention (MNT4P) program, and by private donations. There were 204 people in attendance at the virtual summit, including approximately 60% patients or family members, and 40% clinical providers.

Hans Andersson, MD, of Tulane University gave an overview of MCAD deficiency, focusing on:

- The biochemistry of MCADD and the pathophysiologic mechanisms of the disorder
- Newborn screening and its impact on outcomes
- Treatment and management of MCADD
- Strategies for ensuring healthy outcomes

Rani H. Singh, PhD, RD, LD, of Emory University presented on:

- Nutritional management of MCADD
- Results of the pre-summit survey of registrants
- Research updates (newborn screening, management principles, carnitine supplementation and lived experiences of patients and families)
- Clinical pearls of wisdom regarding acute management
- Patient perspectives

The speakers then answered questions from the attendees, which included:

- MCADD and COVID
- Anti-nausea medication usage
- Recommendations for managing diarrhea
- Use of glucometers
- Knowing the symptoms of hypoglycemia
- Issues related to rural hospitals
- Recommendations for surgeries and procedures
- Cardiac evaluations

A panel consisting of mothers of children with MCADD and an adult with MCADD was moderated by Angela Wittenauer, MSN, FNP-C, RN, a nurse practitioner at Emory University Department of Human Genetics.

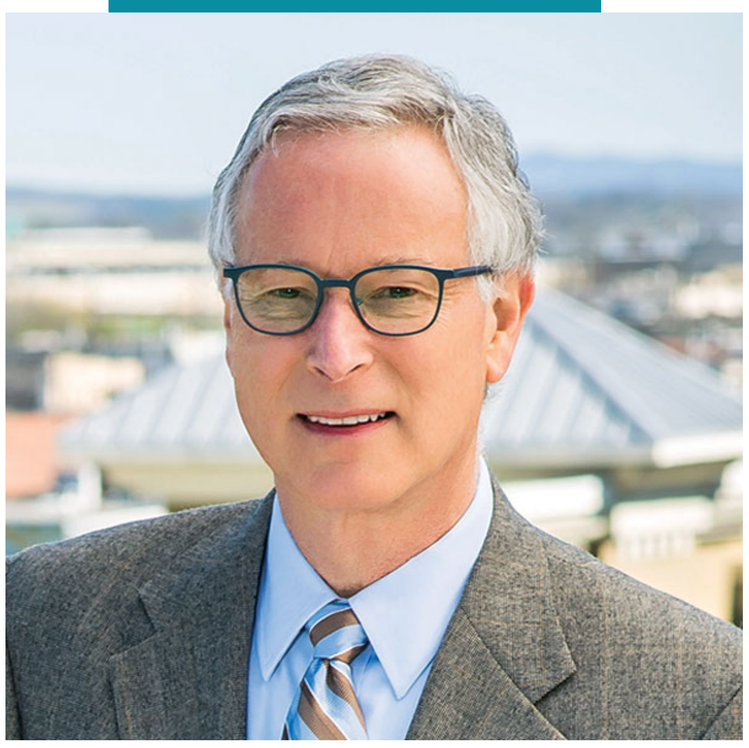
- Experiences with diagnosis and management
- Advocacy experiences
- Sharing information with primary care providers, teachers, etc.
- Experiences with rural emergency departments
- Anxieties
- Self-care
- Health issues in adults with MCADD

Three video presentations are available online: Introduction, Medical Commentary, and Nutritional Management. If you watch the videos, please complete the feedback survey afterwards here.

MEET SOME OF OUR REGIONAL GENE-ies!

RDAC LEADERS: Council Chairs of Rare Disease Advisory Councils of the SERN States

ALABAMA



BRUCE R. KORF, MD

University of Alabama at Birmingham, School of Medicine - The Alabama RDAC was created by the Alabama State Legislature in 2017. I had been contacted by several patient advocates prior to that time asking for my help in formulating the proposal to the legislature. Once the RDAC was approved I was asked to serve as the inaugural chair, and then helped to identify others to serve as members. The RDAC is interested in helping individuals with all forms of rare disease. We have several patient advocates as well as clinicians and researchers working on various types of rare disorders on the RDAC. My personal areas of focus are specific neurogenetic conditions (neurofibromatosis, tuberous sclerosis complex, von Hippel Lindau syndrome), and I run an Undiagnosed Diseases Program at UAB.

FLORIDA

MELISSA JORDAN, MS, MPH

Florida Department of Health - As the Chair of Florida's Rare Disease Advisory Council (RDAC), I have the pleasure of working with a committed group of stakeholders, including individuals and caregivers of loved ones with rare diseases, state agencies, health care providers, researchers, advocacy groups, and insurance and pharmaceutical industries. The RDAC was established in Florida in July 2021 to improve health outcomes and quality of life for individuals in the state who have rare diseases. Together, RDAC works to develop recommendations to improve treatment of rare diseases. I am excited that Florida's RDAC will continue to be on the cutting edge of research protocols and treatments for Floridians living with rare diseases.

GEORGIA

BETH NGUYEN, RN

Rare STRIDES, NORD GA - From 2014 to now, I led Annual Rare Disease Day gatherings in Liberty Square and inside the Georgia Capitol, served as a key advisor to draft the legislation for the Georgia RDAC, met with legislators in Georgia and in Washington DC, testified to the Georgia General Assembly, and led efforts to establish the Georgia RDAC that passed into law. My dual journey as a registered nurse with a rare disease opened my eyes to the complex challenges rare patients face when they seek medical care and inspired a fierce passion within to serve my rare community. Patients with rare diseases deserve excellence in healthcare to prevent complications and help save lives!

SERN FAMILY ALLIANCE ACTIVITIES

The SERN Family Alliance, formerly known as the Consumer Alliance, is a group of individuals with genetic conditions and their family members who work with SERN to provide their perspective on healthcare services for individuals with genetic conditions. The SERN Family Alliance is now under the leadership of Frances Beasley and Linda Starnes. Frances is the parent of a young adult who was diagnosed with a urea cycle defect at the age of two. She has been actively involved with SERN for over 10 years and serves on the Family Center Advisory Committee. She is the Chief Development Officer at Victory Junction – a year-round camp for children living with serious illnesses and chronic medical conditions. Linda brings to the table her experience with family engagement, partnership, leadership, and advocacy through over twenty years working on various state and national boards related to health care, public health, disability, and higher ed, including the SERN Family Alliance for more than ten years. Her two adult children brought her to this work due to their having different rare conditions (Sotos Syndrome and Congenital Bilateral Perisylvian Syndrome). Currently Linda is the Statewide Family Leader for the Florida Department of Health's Title V Children and Youth with Special Health Care Needs Program, and she serves on the Family Center's DEI Workgroup and the NCC's Healthcare Access and Financing Workgroup.

In addition to providing the patient and family perspective to various SERN activities, the SERN Family Alliance has also undertaken projects of their own. They developed Emergency Preparedness Toolkits for a variety of conditions, that are available in both English and Spanish. They are currently exploring ideas for their next project. All adult individuals with genetic conditions, as well as family members of individuals with genetic conditions, in the southeast region are invited to participate in the SERN Family Alliance. If you are interested in joining the SERN Family Alliance, please email serngenetics@emory.edu.

SERN/SERGG 2023 ANNUAL MEETING

July 13-15, 2023

Charleston Marriott Hotel

Charleston, SC

Register Here

HEALTHCARE INTERPRETER TRAINING FOR PEDIATRIC GENETICS

Healthcare interpreters from the SERN region will be participating in a seven-hour virtual training (led by nationally recognized trainer Cynthia E. Roat, MPH) to help them better understand and more accurately interpret the information provided during a genetics appointment. The training is hosted by SERN and HRSA's National Coordinating Center for the Regional Genetics Networks (NCC), and will be held over two consecutive Saturdays in April.

NCC also provides genetics-related resources for healthcare interpreters, including glossaries of common genetics terms in Arabic, Chinese, French, Korean, Polish, Portuguese, Spanish, Tagalog and Vietnamese.

RECENT RESEARCH ON TELEGENETICS

Jessica Corcoran, Caitlin Marley Campbell, and Sigrid Ladores from the University of Alabama at Birmingham School of Nursing recently published a study of the impact of telemedicine on healthcare visits related to cystic fibrosis (CF). They conducted qualitative, semi-structured interviews as part of a larger mixed-methods study examining the current state of fertility preservation counseling for women with CF. Included in the interviews were partners of women with CF as well as CF healthcare providers. The interviews were conducted between September 2019 and July 2020, before and during the national shutdown due to COVID-19 and consequent increase in use of telemedicine. The authors found that participants commented on their experience with telemedicine and its utility for patients and their families. Themes related to telemedicine that arose from the interviews included:

· **'increased connection between healthcare team and family' -** Partners of patients with CF reported that telemedicine provided more opportunities for them to participate in healthcare visits.

· **'increased efficiency of healthcare appointments' -** There is decreased burden on families, by removing the barriers of traffic and childcare needs.

· **'improved interdisciplinary collaboration' -** Both partners and providers commented on the ability to easily include multiple providers in telemedicine-based visits.

Corcoran J, Marley Campbell C, Ladores S (2023) Transitioning to telehealth during the coronavirus disease 2019 pandemic: Perspectives from partners of women with cystic fibrosis and healthcare providers. Chronic Illness 19(1):95-101.

LOW PROTEIN RECIPE

ZUCCHINI BREAD

INGREDIENTS

- 1 1/2 cups Zucchini, grated
- 3/4 cups Sugar, granulated
- 1/4 cup Brown Sugar
- 1/2 cup Vegetable Oil
- 1/2 Cup Water
- 1 tsp Vanilla Extract
- 3 Cups Cambrooke MixQuick
- 1/2 tsp Salt
- 2 tsp Ground Cinnamon
- 1 tsp Ground Nutmeg

INSTRUCTIONS

- Preheat oven to 350 degrees Fahrenheit
- Grease and flour 1 large loaf pan/ a 9x13 baking dish/ small-medium bundt cake pan
- Combine the first 6 ingredients (zucchini -- vanilla) in a large mixing bowl
- In a separate bowl, combine the remaining 4 ingredients (baking mix -- nutmeg), and mix well with a wire whisk
- Add powdered mixture to wet mixture, mixing until completely combined
- Pour into prepared pan and bake 30-45 minutes
  - NOTE: Watch carefully, as the time depends on the type of pan used. Bread is done when an inserted toothpick comes out clean.
- Cool 10 minutes in pan.
- Loosen sides of bread with a knife and flip over on wire rack to remove from pan
- Cool completely (at least 2 hours) before cutting

ADDITIONAL INFORMATION

Yield: 12 Slices
Serving Size: 1 Slice

Nutrition Facts (Per 1 Slice):
PHE: 1.2 mg
LEU: 2.1 mg
TYR: 0.58 mg
Protein: 0.13 g
Calories: 252

Nutritional analysis conducted by RDs at Emory University.

UPCOMING EVENTS

- Healthcare Interpreter Training – April 16 and 23
- Florida Family Leader Network (FFLN) webinar *"Engaging Young Adult Patients in Advocacy."* For more information, contact the FFLN and request to be added to their email listserv – April 20
- Georgia Association of Genetic Counselors Annual Meeting - April 28
- Secretary's Advisory Committee on Heritable Disorders in Newborns and Children Meeting – May 4-5
- NORD's Living Rare, Living Stronger Patient and Family Forum – May 6
- Florida Family Leader Network (FFLN) webinar *"The Day the Dinosaurs Came to Town: Living Through a Disaster"*, with two family leaders in Florida who dealt with living through the devastation of Hurricane Ian last fall. For more information, contact the FFLN and request to be added to their email listserv. – May 18
- Florida Association of Genetic Counselors Annual Educational Conference - May 18-19
- Metabolic Camp - June 19-24· SERN/SERGG Annual Meeting – July 13-15
- National Genetic Education and Family Support Center is planning a series of three spring webinars developed by the DEI workgroup – dates to be announced.

AWARENESS CALENDAR

April

- **Fabry Disease Awareness Month** with the National Fabry Disease Association
- Georgia Association of Genetic Counselors Meeting (April 28)
- **International Pompe Day** (April 15) with the International Pompe Association
- **National DNA Day** (April 25)
- **National Minority Health Month** with U.S. Department of Health and Human Services Office of Minority Health

May

- **Osteogenesis Imperfecta Awareness Week -** (May 6 (Wishbone Day) -- May 13) with the Osteogenesis Imperfecta Foundation
- Public Health Genetics Week (May 15-19)
- **Huntington Disease Awareness Month** with the Huntington's Disease Society of America
- **Cystic Fibrosis Awareness Month** with the Cystic Fibrosis Foundation
- **Neurofibromatosis Awareness Month** with the Children's Tumor Foundation

June

- **World Sickle Cell Day** (June 19) with the Sickle Cell Disease Association of America

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