SERN SOUTHEAST REGIONAL GENETICS NETWORK

JANUARY - MARCH 2022 NEWSLETTER

TELEMEDICINE NEWS

DR. MIKE LYONS AWARD: GGC'S LYONS HONORED AS A TELEHEALTH PIONEER

Congratulations to Michael Lyons, MD, Director of Clinical Services at the Greenwood Genetic Center (GGC), who has been named the 2021 Telehealth Pioneer by Palmetto Care Connections. The award was presented at the organization's 9th Annual Telehealth Summit of South Carolina on November 10th. The statewide award recognizes an individual champion in the areas of clinical care, education, or policy who implemented a telehealth program.

Read more about the award on our website.

GENETICS ECHO PROGRAMS

Project ECHO (Extended Community Health Outcomes) is a provider training method developed by Dr. Sanjeev Arora at the University of New Mexico to help eliminate health disparities by ensuring that the right knowledge is in the right place at the right time. Project ECHO has an "all teach, all learn" approach that incorporates both didactic learning and interactive case-based learning. Learners contribute by presenting and discussing cases, with input from expert practitioners. Genetics-based ECHO programs in the SERN region include:

- The University of Mississippi Medical Center hosts the Pediatric Genetics ECHO program, which aims to help primary care providers improve their knowledge and confidence in their ability to identify, evaluate, and manage individuals with genetic conditions.

- Emory University will soon be presenting the Georgia Genetics in Pediatrics Practice ECHO training program to help primary care providers connect with and learn from regional genetics experts. For more information, contact sernegenetics@emory.edu.

- Emory University also hosts the Genetic Nutrition ECHO Traineeships. These 12-session traineeships have been created to help fill the gap of metabolic specialty training among metabolic dietitians and other clinical providers of patients with inherited metabolic disorders. Genetic Nutrition ECHO staff are currently enrolling the next small cohort of healthcare providers from the US and internationally to begin in February 2022. For more information, contact eGNA@emory.edu.

MEET REGIONAL GENE-ies!

IN HONOR OF MEDICAL GENETICS AWARENESS WEEK, WE ARE HIGHLIGHTING MEDICAL GENETICISTS FROM THE REGION

ALABAMA

Dr. Joy Dean, Institution: University of Alabama at Birmingham, Specialty: Pediatric Genetics and Genomics - My favorite part of my job is having the opportunity to form long term relationships with my patients and their families. From initially meeting with families to discuss abnormal newborn screen results to following-up regularly with these patients throughout their lifespan, you are able to form a strong bond with each family.

FLORIDA

Dr. Robert Zori, Institution: University of Florida at Gainesville, Specialty: Pediatric Genetics and Metabolism - Genetics has allowed me to not limit myself to patients of a certain age or problem, but instead concentrate on the patient in front of me irrespective of what their needs are, applying all the tools of medicine and especially genetics including clinical, biochemical, chromosomal, or molecular.

GEORGIA

Dr. Rossana Sanchez, Institution: Emory University, Specialty: Clinical & Biochemical Genetics - Clinically, genetics allows me to aid with the diagnosis and clinical management of patients and offer support to their families. In education, I help bring awareness of the field of genetics to all generations and teach on how to diagnose and test for these. In research, I am able to participate in clinical trials and aid in development of new therapies for rare disorders, most of which still lack approved treatments.

LOUISIANA

Dr. Jariva Upadia, Institution: Tulane University School of Medicine, Specialty: Clinical Genetics - I strive to provide quality of care for my patients with the most current knowledge and technologies. As a clinical geneticist, my main area of interest in the diagnosis and treatment of in inborn error of metabolism.

MISSISSIPPI

Dr. Kirmse, Institution: University of Mississippi Medical Center, Specialty: Clinical Biochemical ("Metabolic") Genetics - I am a pediatrician who takes care of kids with rare disorders, in particular rare genetic disorders that affect energy metabolism. My favorite part of the job is still patient care - getting answers for the patients and their families and helping them stay healthy through multidisciplinary team-based medicine.

NORTH CAROLINA

Dr. Chad Haldeman-Englert, President of NC Medical Genetics Association Institution: Mission Fullerton Genetics Center, Specialty: Clinical Genetics - I have definitely found the right career for me. I really enjoy the daily interactions with patients and their families, but I have also become interested in how we can use genetics to make us healthier. Our field is so fascinating, and I look forward to its future!

SOUTH CAROLINA

Dr. Neena Champaigne, President of SERGG; Institution: Greenwood Genetics Center, Specialty: Clinical Genetics - Dr. Champaigne provides clinical evaluation, care and counseling for metabolic patients across the Upstate, with clinics in both Greenville and Greenwood. She also provides short- and long-term follow-up for the South Carolina Newborn Screening Program.

TENNESSEE

Dr. J Austin Hamm, Institution: East Tennessee Children's Hospital, Specialty: Pediatric Genetics - I chose a career in medical genetics because I find it gratifying to care for patients with rare diseases. There are limited resources available to help patients and families struggling with rare disease and I find it personally and professionally rewarding to be able to provide patients with diagnostic and therapeutic options for an increasing number of diseases.

NORD RARE DISEASE CENTER OF EXCELLENCE AWARDEES IN THE SERN REGION

In November, 2021, the National Organization for Rare Disorders (NORD) announced 31 NORD Rare Disease Centers of Excellence across the United States. The goal of the network is to expand access, advance care and enhance research for patients with rare diseases. According to the National Institutes of Health, any disease that affects fewer than 200,000 people in the United States is a rare disease. With over 7,000 known rare diseases, approximately 25 million to 30 million Americans live with a rare disease. More than 90% of rare diseases lack FDA-approved treatments.

Six institutions in the SERN region have been designated as NORD Rare Disease Centers of Excellence:

- Duke Health Rare Disease Center
- Emory University and Children's Healthcare of Atlanta
- UNC Children's – North Carolina Children's Hospital
- University of Alabama at Birmingham Medicine/Children's of Alabama
- University of Miami Miller School of Medicine
- Vanderbilt University Medical Center

SERN FUNDED RESEARCH

NBS-PKU Connect was a web-based self-report patient registry supported by SERN that collected information on current diagnosis, management, and treatment practices from respondents with PKU or hyperphenylalaninemia, or their caregivers. Drs. Kenneson and Singh recently published an analysis of data from 219 individuals with PKU or hyperphenylalanemia from the US who completed the natural history survey. Key findings from the analysis include:

- Despite recommendations to maintain blood phenylalanine concentrations in the therapeutic range throughout life, it is not uncommon for adults with PKU to discontinue dietary management of their disorder

- One-quarter of respondents reported psychological co-morbidities, with anxiety and ADD/ADHD being the most common.

- The incidence of psychological co-morbidities increased with age, and individuals who had been off diet at some point in their lives had a higher chance of having a psychological co-morbidity.

- Special education or other special services were more likely to be reported by individuals who were diagnosed after one week of age

Read the complete article here.

UPCOMING EVENTS

JANUARY

Lunch and Learn
- Date: January 28, 2022
- Topic: GAMT deficiency
- Registration open now

FEBRUARY & MARCH

eGNA
- Date: February 25th & March 4th, 2022
- Topic: Tyrosinemia (HT1)

SAVE THE DATE

SERN Healthcare Interpreter Training
- Date: April 16th & 23rd, 2022
- Registration opens January 13th
- Visit the SERN website for more information

SERN / SERGG annual meeting
- Date / time: July 14-16, 2022
- Location: Asheville, NC
- Keynote speakers: SERN & SERGG

Watch out the call for abstracts - some student awards will be made

SUBMIT YOUR EVENTS TO BE POSTED ON THE SERN WEBSITE!

LOW PROTEIN RECIPE: PATRICK'S IRISH MINESTRONE

INGREDIENTS

- 1/3 cup canola oil
- 4 medium garlic cloves, minced
- 400 grams onion, chopped
- 170 grams celery, chopped
- 20 grams fresh flat leaf parsley, chopped
- 170 grams tomato paste
- 10 cups vegetable broth, canned
- 90 grams cabbage, chopped
- 155 grams carrots, peeled and thinly sliced
- 2 tsp salt
- 1/4 tsp pepper
- 1/8 tsp sage, dried
- 1/8 tsp basil, dried
- 140 grams zucchini, halved lengthwise, cut into 1/2 inch pieces
- 120 grams green beans, cut into 1-inch pieces
- 1/2 cup low protein Ditalini Pasta

INSTRUCTIONS

1) Heat oil in stockpot over medium-high heat

2) Add garlic, onion, celery, and parsley. Sauté until the vegetables soften and the onion is translucent, about 4 minutes

3) Stir in the tomato paste, broth, cabbage, carrots, salt, pepper, sage, thyme, and basil

4) Mix well and bring to a boil

5) Lower heat, cover and simmer for 35 minutes

6) Add zucchini and green beans

7) Add pasta, bring to a boil, reduce heat to medium-low, and cook for 10-15 minutes more until pasta is tender

EXTRA INFORMATION

- Yields 12 1/2 cups
- Serving size: 1/2 cup

Nutritional information (per 1/2 cup serving)
- Calories: 56
- Protein: 0.85 grams
- Phenylalanine: 27 mg
- Leucine: 35 mg
- Tyrosine: 0.016 g

Recipe from: Apples to Zucchini: A Collection of Favorite Low Protein Recipes by Virginia Schuett & Dorothy Corry

AWARENESS CALENDAR

JANUARY:

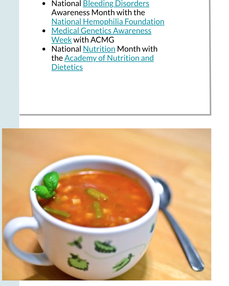
- Happy 2022!
- National Birth Defects Prevention Month with the National Birth Defects Prevention Network

FEBRUARY:

- Rare Disease Day 2022 (Feb. 28) with multiple organizations

MARCH:

- National Bleeding Disorders Awareness Month with the National Hemophilia Foundation
- Medical Genetics Awareness Week with ACMG
- National Nutrition Month with the Academy of Nutrition and Dietetics



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