



OCTOBER - DECEMBER 2023 NEWSLETTER

Closing of HRSA's Regional Genetics Networks

We recently received communication from HRSA that SERN and the other regional genetics networks will no longer be funded after this current grant year is completed on May 31, 2024, after nearly two decades of funding. We will work with our partners such as the [Southeast Regional Genetics Group \(SERGG\)](#), [Genetic Metabolic Dietitians International \(GMDI\)](#), and Emory University's [Medical Nutrition Therapy for Prevention \(MNT4P\)](#) program to ensure that the resources we've developed continue to be available to you. In our future newsletters or other communications, we will share information about where to access our various resources and projects as we plan for the SERN website to come to a close after May 31, 2024.



SERN-FUNDED RESEARCH

Perceptions and Experiences of Families of Infants Diagnosed with X-Linked Adrenoleukodystrophy (X-ALD) via Newborn Screening in the Southern United States

X-linked adrenoleukodystrophy (X-ALD) is a genetic disorder caused by a pathogenic variant in the *ABCD1* gene that results in a variable phenotype ranging from adrenal insufficiency to cerebral demyelination. If caught in the early stages, the cerebral form of X-ALD can be effectively treated. X-ALD was added to the Recommended Uniform Screening Panel in 2016. Early diagnosis of X-ALD via newborn screening allows for monitoring and detection of disease in the earliest stages. To better understand parents' experiences and their feedback for the referral processes, Emory University genetic counseling student Sharanya Iyer conducted qualitative interviews with seven parents/caregivers of infants diagnosed with X-ALD via NBS in the Southern United States.

Parents had both positive and negative feedback at various points in the screening and diagnosis process. Before NBS, which is performed via heel prick, some parents were unaware of the test's purpose. During the initial result call out, parents expressed mixed emotions of confusion, anxiety, and fear. Most parents felt that they lacked an understanding of the condition and did not like the way the NBS result was disclosed. All of them were referred to genetics and had positive feedback on this interaction. Endocrinology and neurology follow-up was required for everyone who tested positive. A few of the barriers include lack of access to clinics during COVID-19 pandemic and lack of providers' knowledge about X-ALD. Overall, parents were grateful for NBS but expressed concerns about the medicalization of their infants' childhoods.

The study results suggest ways to improve the experience of parents whose children are diagnosed with X-ALD through NBS. As more states add X-ALD to their NBS programs, it is important to understand the referral process and potential pitfalls to avoid. Improvements can be made by educating primary care providers, creating more access to education materials, and providing mental health support to parents. This study has been submitted for publication in a peer-reviewed journal.

MEET SOME OF OUR REGIONAL GENE-ies!

GENETIC COUNSELOR TRAINING PROGRAM DIRECTORS

UNIVERSITY OF ALABAMA AT BIRMINGHAM



R. Lynn Holt, MS, CGC
Program Director

One of the biggest differences that I have seen since the establishment of the UAB program in 2007, is the advancement and complexity of genetic testing in the clinical setting as well as professional roles in clinical laboratories. In order to prepare UAB graduates for these advances, the program has expanded the depth of education in these areas throughout the curriculum. We have expanded our faculty to include a laboratory genetic counselor who teaches throughout the program as well as the two-week intensive course in the summer term entitled "Clinical Laboratory Rotation". During this course, students gain in-depth experience in biochemical, molecular and cytogenetic laboratory protocols, reporting and genetic counseling roles.



Jessica Denton, PhD, MS, LCGC
Associate Program Director

Despite the changes in technology and advances in genetics, the most important skill for genetic counselor remains our quality counseling skills. Quality counseling skills require critical thinking, compassion, self-awareness, active listening, and the ability to explain complicated information often in a time of crisis. We foster this development in the UAB program with the inclusion of counseling course work from the UAB Counseling graduate program and communication-based medical simulations, role plays and reflective practice as part of the genetic counseling curriculum in addition to skill demonstration during clinical rotations to help students identify their personal counseling style.

EMORY UNIVERSITY

The Focus Internship is a unique part of the Emory Genetic Counseling Training Program (GCTP) curriculum which occurs over the entire 21 months of the program. During the focus internship, students develop a research project in collaboration with a mentor and participate in a variety of activities in their mentor's specialty area. Focus internships are available across several clinical specialties as well as public health and education.

There are not enough genetics professionals to serve the population in need of our services. It is therefore essential for genetic counselors to develop interprofessional collaborations. At Emory, there are many interprofessional activities where genetic counseling students have the opportunity to work with students in medicine, physical therapy, physician assistant, nursing, anesthesia assistant, medical imaging, and/or nursing. These activities build a culture of respect and collaboration that will extend to their future workplaces.

In such a fast-moving field, graduates will inevitably encounter new technologies and situations that they were exposed to in graduate school. Emory GCTP students complete ten rotations, which encompass both well-established specialties as well as those that are newly developing. These experiences empower graduates to bring the profession to new places, while also providing them with a network of mentors beyond graduate school.



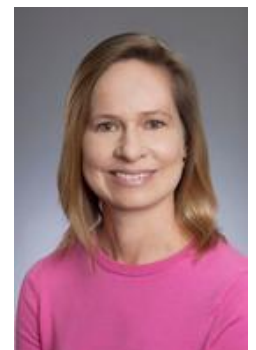
Lauren Lichten, MS, CGC
Program Director



Ami Rosen, MS, CGC
Associate
Program Director



Nadia Ali, PhD
Assistant
Program Director



Christine Stanislaw, MS, CGC
Director of
Student Rotations

MEET SOME OF OUR REGIONAL GENE-ies!

GENETIC COUNSELOR TRAINING PROGRAM DIRECTORS

UNIVERSITY OF NORTH CAROLINA GREENSBORO



Lauren E. Doyle, MGC, CGC
Program Director



Rachel Mills, MS, CGC
Capstone & Research
Coordinator



Randi Stewart, MS, CGC
Assistant Program Director

The MS Genetic Counseling program at the University of North Carolina Greensboro (UNCG) takes a holistic approach to genetic counseling education. The program values integrating concepts during training with an emphasis on cultural humility and responsiveness to best serve patients and families. Community engagement within and beyond the training program has been a proud hallmark of the program for over 20 years, and students have expressed the value of the sense of community and belonging in their training experience.

Much like the process of genetic counseling itself, the program builds content and skills throughout the course of training while also tailoring as much of the process as possible to the individual student. With an emphasis on personal and professional development throughout the program, UNCG carries the holistic focus on genetic counseling to students during training as well as to patients and the field.

MEDICAL UNIVERSITY OF SOUTH CAROLINA

The Master of Science in Genetic Counseling (MSGC) program at the Medical University of South Carolina (MUSC) is thrilled to have 14 students in our inaugural cohort, after receiving ACGC New Program Accreditation. MUSC is the only academic medical center in South Carolina, and we are the first in the Southeast region to offer a hybrid program with coursework delivered in an online and asynchronous format. In fact, most students will not be in Charleston, SC, in-person until clinical rotations begin next summer, allowing year one to be completed fully remote. The program adopted this modern model to make the genetic counseling profession more accessible and convenient to all types of learners. We also accept a larger than average class size. The workforce demand for genetic counselors continues to increase, and there are consistently over three applicants per training spot nationally, so we strive to optimize resources to train as many qualified students as possible and meet workforce needs of the future. Through strategic initiatives, the MUSC Health clinical genetics workforce has also grown, with 15 genetic counselors now providing services across prenatal, oncology, pediatrics, cardiology, neurology and pulmonary practice areas. While a majority of rotation placements for our students will be at MUSC, we also look forward to partnership with Greenwood Genetic Center and other institutions to provide a comprehensive and balanced training experience for all students.



Kimberly Foil, MS, CGC
Program Director



Libby H. Malphrus, MS, CGC
Assistant Program Director

MEET SOME OF OUR REGIONAL GENE-ies!

GENETIC COUNSELOR TRAINING PROGRAM DIRECTORS

UNIVERSITY OF SOUTH CAROLINA



Whitney Dobek, MS, CGC
Program Director

I always sum up the strength of the University of South Carolina (USC) Genetic Counseling program (GCP) in one word: community. But we cultivate this strength in more than one way. Upon arrival in Columbia, our students become a part of an amazing community of current students, faculty, local genetics professionals, and growing network of alumni that make the USC GCP what it is. Additionally, participating in community outreach is a key component of our curriculum. These engagement experiences range from education at local schools to volunteering at fundraising and awareness events. We value supporting local advocacy groups at the heart of where genetics intersects with the patient population we serve. Educating our community, fostering a growth in genetics knowledge, and increasing awareness of genetic counseling will be a piece of all genetic counselors' careers. Community in all senses is a critical part of becoming a successful genetics professional.



Amy Wardyn, MS, CGC
Assistant Director,
Thesis Research



Jessica Fairey, MS, CGC
Assistant Director,
Fieldwork

VANDERBILT UNIVERSITY

Vanderbilt School of Medicine Master of Genetic Counseling has a curriculum that is integrated to ensure our learners receive meaningful, rigorous, and intentional opportunities to gain knowledge and skills in the field of genetic counseling. Our courses were designed by genetic counselors and are taught by genetic counselors. In the first year, every week wraps up with a clinical case discussion called our "Integrate Case." This experience is student driven and allows the application of the week's lessons.

Increasing the diversity of our field is essential to serving all families impacted by genetic disease. We are partnering with the Tennessee Genetic Counselor Association to increase awareness of the field at schools with underrepresented groups in medicine. We weave cultural competency, and antiracism training throughout all our courses and have opportunities for faculty development in anti-racism and inclusive teaching practices.

Building a self-reflective practice is an essential skill for all genetic counselors. Being self-reflective means you take notice every day how you are taking care of yourself first and then how you are caring for others. You are aware of your well-being and the impact you are making on those around you. You recognize when you can learn more, act more and do better. You make a plan to achieve those goals, striving for self-improvement and improved well-being. These practices are woven into our curriculum and our students have a dedicated course, "Building a Self-reflective Practice", when they have a regular time for cohort support and contemplation.



Martha Dudek, MS, CGC
Program Director



Jill Slamon, MA, MS, CGC
Assistant Program Director

MEET SOME OF OUR REGIONAL GENE-ies!

GENETIC COUNSELOR TRAINING PROGRAM DIRECTORS

WAKE FOREST



Emily Lisi, MS, CGC
Program Director



Lauren Baldwin, MS, CGC
Assistant Program Director

The Wake Forest Genetic Counseling Program has New Program Accreditation through ACGC and graduated its first cohort in 2023! Our program is unique in several ways: our curriculum places emphasis on ethics issues that arise in genetic counseling as well as health disparities affecting our populations. We also offer two tracks for our capstone research experience - a traditional clinical research track as well as a community outreach track in which students can meet an educational or advocacy need in the community and write a paper reflecting on their experience.

I feel like the biggest challenge in genetic/genomic medicine today is the way that integration of new technologies is increasing already existing health disparities in the provision of genetics healthcare. Underserved populations are being further marginalized by the use of genomic data for medical decision-making with little representation from these populations. The students at Wake Forest are introduced to these concepts both inside and outside of the classroom and are encouraged to be advocates for the marginalized communities through clinical and research activities.

Genetic counselors in the current healthcare climate must be flexible and adaptable while also remaining thoughtful stewards of our role as patient advocates. The landscape of genetic counseling and testing is changing daily- genetic counselors must be able to discern the potential ramifications of new technologies for our patients and empower them to make decisions that reflect their own values and beliefs. Our curriculum allows students to develop an understanding of both the clinical application of emerging technologies as well as their ethical implications in modern society.

UNIVERSITY OF SOUTH FLORIDA

Our program is unique because we are an MSPH (Master of Science in Public Health) program with a concentration in genetic counseling. We meet all the same criteria of an accredited Genetic Counseling program and also provide additional training in public health. We have also set up a spiral curriculum and start clinical rotations in the first year. We are also one of the most affordable genetic counseling graduate programs in the U.S.

There are many challenges facing the profession. One of these is to demonstrate genetic counselors' added value as part of the healthcare team and how we contribute to patient outcomes and quality service delivery. In our program, we try to get students to think critically about what they do and which parts are contributing most to improving patient experiences and outcomes.

I believe that critical thinking and ability to wade through vast amounts of information and picking out what is most important and what information can be trusted are critical skills. We do a lot of problem-based/case-based learning to develop these skills.



Deborah Cragun, PhD, MS, CGC
Program Director



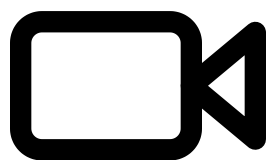
Nevena Krstić, MS, CGC
Associate Director

Chatbots for Pre-Test Education: Cleveland Clinic Experience

At the 2023 SERN/SERGG Annual Regional Genetics Meeting, Dr. David Flannery from the Cleveland Clinic presented on the use of chatbots for pre-test education related to genetic testing. Chatbots are one tool that can be used to address the challenges of the limited genetics workforce, as chatbots can take on some of the repetitive and scriptable tasks, thus freeing up time for genetic counselors to focus on more nuanced and difficult aspects of care.

In a recent [publication](#) by Al-Hilli and colleagues at the Cleveland Clinic, a randomized study which compared the effectiveness of pre-test education using a chatbot versus in-person genetic counseling for women who were newly diagnosed with breast cancer found no difference between the groups in satisfaction or understanding.

Dr. Flannery's presentation resulted in a lively discussion. Participants from the audience also reported that chatbots are used at other sites for the return of negative genetic test results. Issues raised by the participants included the impact of genetic counseling licensure laws on the ability to offer some elements of counseling by chatbot.



Videos of talks from the SERN portion of the SERN/SERGG 2023 Annual Genetics Meeting are now available on our YouTube Channel.

Click [here](#)



SAVE THE DATE

SOUTHEASTERN REGIONAL GENETICS GROUP ANNUAL MEETING

July 11-13, 2024

Renaissance Marriott Hotel
Asheville, North Carolina



Metabolic Camp is turning 30!

The 30th Metabolic Camp for girls and women with the rare disorders of phenylketonuria (PKU) and maple syrup urine disease (MSUD) will be held on Emory University Campus on June 10-15, 2024.

Please watch the Metabolic Camp [website](#) for registration information as we get closer to the date of the camp.

MORE HIGHLIGHTS FROM THE 2023 SERN/SERGG ANNUAL MEETING: SERN-FUNDED RESEARCH

Women with Phenylketonuria Report that Health Management Education and Social Support at Emory Genetics Metabolic Camp Prepared Them for Later Pregnancy

by Teresa D. Douglas, Ashley Wrushen, Rosalynn Blair, Rani H. Singh

Women with phenylketonuria (PKU) must maintain a strict low-protein therapeutic diet during pregnancy and maintain blood phenylalanine (phe) levels below 360 μ moles/L to prevent severe complications in the newborn known as Maternal PKU Syndrome. Adherence to the complex dietary and healthcare regimen necessary throughout the full pregnancy is strenuous for most women with PKU. Emory Genetics Annual Metabolic Camp provides a supportive environment for females with PKU to learn about the impact of nutrition and dietary self-management on health and birth outcomes so they can effectively self-manage their medical diet and metabolic health needs throughout life, including during pregnancy.

In this study, camp alumnae were invited to complete a 16-question survey with questions about how their first pregnancy experience was influenced by various interactions, activities, and educational components from prior attended camps. Participants could also opt-in to a half-hour 17-question interview with open-ended questions about whether the learning and social components of metabolic camp influenced their first pregnancy experience, and interviewees were given an opportunity to impart recommendations relevant to future camps and campers. Study participants described camp learning activities and social components that helped them prepare for their first pregnancy. Data collection for this study is ongoing.

Interview quotes from camp alumnae

"They explained the importance during your pregnancy to keep a food record. Told that weekly blood phe needs to be done during pregnancy...I learned how to prick my own finger...knew I could do it and not be hurt by it."

"It's fun, you get the support system, you find other girls. Cause half the time there is no one around. So you feel alone your whole life with it...then you go to camp and get a support system of sisters who have it, know what you are going through."

"I go there for a purpose. I take it very seriously. I talk about it all the time. I recommend it highly. That's where I learned about formula, different recipes, how to take blood. I talk to my camp-friends a lot and we are very close."

Qualitative Assessment of Primary Care Providers' Attitudes Towards Genetics Services and Genetics Education

by Aileen Kenneson, Yasmin Thornton, Chelsea Cole, Rani H. Singh

Primary care providers (PCPs) are considered the "gatekeepers" of genetic services, as they are often responsible for recognizing the patient's need for referral to genetic services. The evolving role of PCPs is unclear and unsupported, with PCPs commonly reporting a lack of knowledge about basic genetic concepts, the availability of genetic services and testing, interpreting genetic test results, and how to refer to genetic services. We conducted interviews with nine PCPs practicing in states in the SERN region about their perceptions of the role of PCPs in genetics and the educational needs of PCPs regarding genetic referrals and genetic testing. Thematic analysis was conducted on interview transcripts.

Themes:

- **Perceptions of genetics:** *Cancer, Family History, Complicated*
- **PCPs' roles in genetics:** *Referrals, Patient/Family Education*
- **Common reasons for referrals to genetics:** *Cancer, cascade testing, developmental delay*
- **Barriers to referrals to genetics services:** *Logistics, lack of knowledge, patient fear, insurance issues*
- **PCPs' educational needs regarding genetics:** *How/when to refer to genetics, test results interpretation, basics of genetics, how to co-manage patients with genetic conditions*

LOW PROTEIN RECIPE

Maple-Roasted Squash

Ingredients:

- 2 pounds Butternut squash, peeled & cubed
- 1 tablespoon Olive oil
- ¼ teaspoon Salt
- ⅓ cup Maple Syrup
- ½ teaspoon Pumpkin Pie Spice
- ⅛ teaspoon Cayenne Pepper

Steps:

- Heat oven to 425°F.
- Line a sheet pan with aluminum foil.
- Place the cubed squash in the pan.
- Drizzle with olive oil, sprinkle with salt and toss to combine.
- Roast squash for 15 minutes
- Meanwhile, stir the maple syrup with pumpkin pie spice and cayenne pepper until combined.
- Drizzle the maple syrup mixture over the squash; toss to coat.
- Roast the squash for another 15-20 minutes or until fork-tender. Spoon the squash, along with the with the pan juices, into a serving dish.

ADDITIONAL INFORMATION

Yield: 4 ½ cups
Serving Size: ½ cup

Nutrition Information: (per ½ cup serving)

Calories: 80
Protein: 1.4 g
Phe: 60 mg (4 exchanges)
Leu: 80 mg (2½ exchanges)
Met: 20 mg:

Recipe from Good Housekeeping Magazine.

Nutritional analysis conducted by RDs at Emory University. Nutritional analysis based on the products used at the time of analysis. Subject to change.

AWARENESS CALENDAR

October

National Down Syndrome Awareness Month with the [National Association for Down Syndrome](#) and the [National Down Syndrome Society](#).

November

National Family Health History Day - November 23 with the [CDC](#)

Genetic Counselor Awareness Day - November 9 with the [National Society of Genetic Counselors](#)



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