# Greenwood Genetic Center

Summer 2023

A Newsletter for the Friends of the Center

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GGC and MUSC partner on Precision Medicine Initiative

# Precision Medicine:

## The Right Treatment for the Right Patient at the Right Time

GGC has embarked upon a formal Precision Medicine Initiative (PMI) with collaboration among divisions across the Center as well as with partners at MUSC.

"Precision medicine involves the use of genetic and genomic information to guide the most effective personalized treatment for each patient," said GGC Director, Steve Skinner, MD. "It's basically finding the right treatment for the right patient at the right time."

GGC's PMI has been designed with a four-pronged approach. "It's what we're calling the Four A's: Access, Analysis, Answers, and Action," added Skinner.

### ACCESS

"If a patient cannot access genetics services it doesn't matter if we have the most effective treatment, or even a cure," said Mike Lyons, MD, GGC's Director of Clinical Services. "It all begins with the removal of any barriers, such as cost, travel, and a shortage of providers, that prevent patients from getting to us."

GGC is working to recruit additional clinical personnel as well as expand the use of technologies to allow patients to be evaluated more quickly.

GGC's recent Duke Endowment grant allowed for the development of eVisits and eConsults which have helped reduce wait times in many of GGC clinics by as much as 60%. The PMI will expand those services to provide further access.

eVisits allow GGC providers and patients to communicate online without the need to wait for an appointment or travel to a GGC office. eConsults are online communications between GGC providers and those in primary care or other specialties. Through eConsults, a patient's primary care provider can ask questions and receive testing recommendations allowing for more rapid access to information and testing.

### ANALYSIS

"While genetic and genomic technologies have been advancing rapidly in recent years, there are still a significant number of patients who lack an answer," said Mike Friez, PhD, Director of Diagnostic Laboratories at GGC. "As amazing as technologies like whole genome sequencing are, there are still disorders that are not detectable by current methodologies."

GGC will investigate a variety of novel technologies to assess the ability of these new assays to identify disorders that have previously been undiagnosable. Using existing patient samples, the Center will develop defined cohorts to evaluate the utility of technologies that go beyond the capability of current tests like whole genome sequencing. These technologies include long-read sequencing, RNA sequencing, and optical genome mapping to detect previously undetectable genetic changes. The development of a GGC data warehouse will also support the PMI by allowing GGC's nearly 50 years of clinical and lab data to be more easily accessible and queried.

"We're looking to identify the next big thing in genetic diagnostics," added Friez. "When genomes don't give us the answer, where should we turn next?"

### ANSWERS

"In order to identify the most effective treatment, we must have an accurate diagnosis," said Rich Steet, PhD, GGC's Director of Research.

GGC has instituted a new Genomic Discovery Program (GDP) to identify undiagnosed patients and use the latest diagnostic and research capabilities to find an answer and understand the mechanism of the condition with the ultimate goal of identifying a treatment.

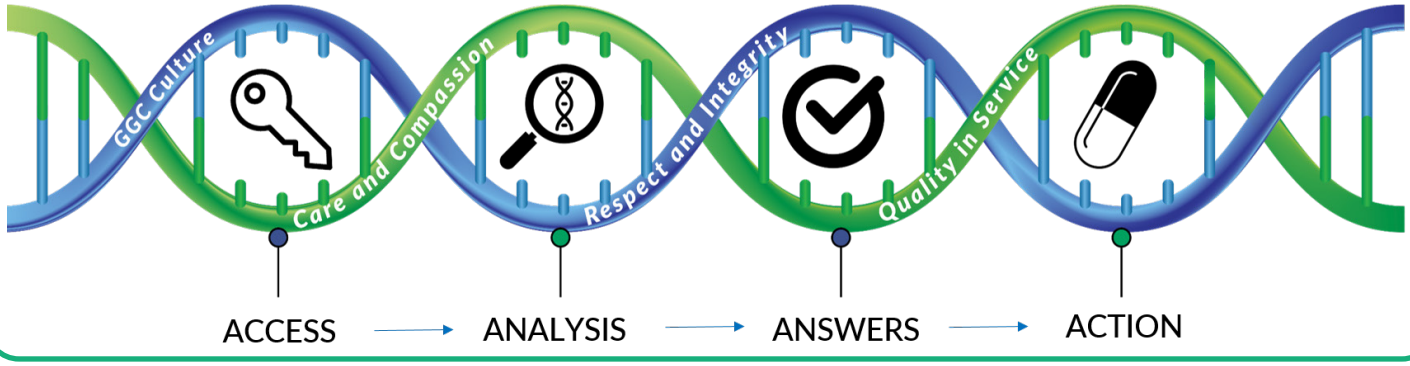
### ACTION

"Seeing patients, analyzing data and finding answers are essential steps, but to fulfill our ultimate responsibility to those we serve, we must be able to act on that information by providing the best and most effective treatments," said Skinner.

To that end, GGC is working to reorganize the clinical trial program under an endowed 'Curry Chair in Genetic Therapeutics.' The Center is also focusing on expanding single patient, or 'n of 1,' clinical trials to advance the ability to treat ultra-rare disorders.

"We are also leveraging the vast expertise and experience that MUSC has developed in clinical trials by enrolling more GGC patients in their clinical trials," added Skinner. "Our affiliation with MUSC will also help us develop drug screening options for patients enrolled in the GDP."

"Through the coordinated and collaborative efforts of this ambitious initiative, GGC is pushing the boundaries of what is currently standard of care in genetics," said Skinner. "We are striving everyday to follow our guiding principles of providing compassionate patient care and the highest quality of state-of-the-art services."



# PRECISION HEALTH PLAN GUIDES GGC-MUSC PARTNERSHIP PRIORITIES

GGC and MUSC Health marked the first anniversary of their membership agreement in May with the development of a five-year strategic plan around precision health.

Precision health uses information about a person's genetic material and its interaction with the environment and lifestyle to prevent, diagnose, and treat diseases more efficiently and effectively. The vision for precision health at GGC and MUSC aims to differentiate the collaborators as a leader in precision health through a three-phase strategic plan.

*Skinner (left) and Cawley at a recent GGC Board of Directors meeting.*

"With the combined internationally-renowned expertise and comprehensive statewide footprint of both GGC and MUSC, this partnership is poised to make precision medicine not only a reality, but the standard of care for all South Carolinians," said Steve Skinner, MD, Director of GGC. "Our Precision Medicine Initiative (p.2) will serve as the basis for a larger, more global joint focus on precision health, which encompasses both personalized patient care for rare genetic disorders as well as broader population-level health initiatives for rare and common conditions with a genetic component."

The strategic plan will lead to the development of a Precision Health Institute to house clinical, research, and educational programs and ensure that all South Carolinians have easy access to genetic testing and understand its application for their health. The first phase of the plan, to be implemented during the upcoming year, will provide for the integration of MUSC Health and GGC operations for a seamless clinical workflow as well as the development of an integrated data warehouse to set the stage for transformational care. Workforce development activities will also be a focus, along with leveraging MUSC Health resources to scale GGC operations.

The joint plan expands upon GGC's four A's of access, analysis, answers, and action with specific goals to improve patient access, diagnostics, and treatments. Projects include the enhanced integration of genetics services and testing between GGC and MUSC, raising awareness of genetic services for patients and providers, expanding testing capabilities, growing clinical trials, and leveraging artificial intelligence and machine learning to support diagnostic endeavors.

"We are thrilled with the progress of this partnership over the past year and look forward to expanding our work together to establish GGC and MUSC Heath as a regional and national leader in population-level genomic technologies and clinical practice standards," said Patrick Cawley, MD, CEO of MUSC Health. "With the implementation of this ambitious program, we will develop a sustainable collaborative model to improve the long-term health and well-being of all citizens of South Carolina."

# SHARE YOUR GGC STORY

How has the Greenwood Genetic Center made a positive impact on your life or the life of your family? Did you finally receive a long-awaited diagnosis? Did a member of our team go above and beyond in caring for your family? We want to hear your story.

As GGC prepares to celebrate its 50th anniversary in 2024, we are developing a video series of patient testimonials, and we would love to include your GGC experience. These brief videos will be highlighted on social media, during events, and on other platforms to raise awareness for the work of the Greenwood Genetic Center.

If you are interested in being a part of this historic project, please let us know by scanning the QR code and completing the interest form or visiting ggc.org/patientstory. We will begin filming this fall and will continue into 2024.

If you have any questions about this project, please reach out to GGC's Director of Communications, Lori Bassett, MS, CGC (lbassett@ggc.org or 864-388-1061) or Clinic Coordinator, Fran Annese, LMSW (fran@ggc.org or 864-941-8160).

*Hadley Wofford, a patient enrolled in the trofinetide trial, is presented with a 'Thank You for being Awesome' trophy by Fran Annese and Dr. Steve Skinner at her final clinical trial visit.*

# A NEW HOPE

## GGC part of trials for first FDA-approved drug for Rett syndrome

Patients and genetics professionals around the globe are celebrating a historic milestone - the first and only FDA-approved medication to treat Rett syndrome, a rare genetic neurodevelopmental disorder.

GGC, an International Rett Syndrome Foundation Center of Excellence, has a long-standing interest and vast expertise in Rett syndrome. As such, GGC was selected to be a clinical trial site for Acadia Pharmaceutical's DAYBUE (trofinetide) which was approved in March for use in adult and pediatric patients with Rett syndrome two years and older.

Rett syndrome affects 6,000-9,000 patients in the US and is caused by a mutation in the *MECP2* gene on the X chromosome. Because of its location on the X chromosome, the disorder mainly impacts females, with males less frequently affected.

“Rett syndrome causes significant and lifelong challenges including developmental regression with the loss of communications skills and purposeful hand use, as well as behavioral and physical symptoms that impact breathing, sleep, growth, and mobility,” said Steve Skinner, MD, Director of GGC and study investigator.

"Prior to DAYBUE's approval, the only available medical interventions were focused on treating individual symptoms of Rett syndrome - seizure meds to treat seizures, GI meds to treat constipation, etc.," said Skinner. "This drug is designed to repair synapses, or connections within the brain, that we hope will lead to a more global improvement of symptoms."

*"We are excited and hopeful for the benefits this will have for our families"*

*-Fran Annese, GGC Clinical Trial Coordinator*

GGC had been involved in earlier safety and efficacy studies of trofinetide, but the Phase 3 clinical trials began just as the COVID pandemic was taking hold.

"It was a huge challenge to conduct a clinical trial in the midst of a global pandemic," shared Fran Annese, LMSW, GGC's Clinical Trial Coordinator. "We had to follow the strict protocols necessary for a successful clinical trial while also taking measures to keep our patients and their families safe."

Despite these challenges, GGC enrolled seven patients with Rett syndrome from across North and South Carolina for the trial. The first part of the study involved a randomized double-blind placebo-controlled study with some patients on the drug and others on placebo. Which patients received the actual drug was unknown to both the investigators and the families. This protocol was followed by an open label study where all enrolled patients received the study drug. Patients visited GGC regularly to receive the drug or placebo and undergo medical exams and other assessments to identify any changes in their condition. Parents and caregivers also completed surveys throughout the study to assess their family member's condition.

The results demonstrated statistically significant improvement in several areas for patients who took trofinetide compared to those taking the placebo.

"This is not a cure," said Skinner, "but many patients are seeing improvements in areas including anxiety, hand movements, and nonverbal communication. And we don't yet know the full benefit of long-term administration of DAYBUE or of starting the drug at an early age when the diagnosis is first made," he added. "That data will take time to assess, but we are very hopeful."

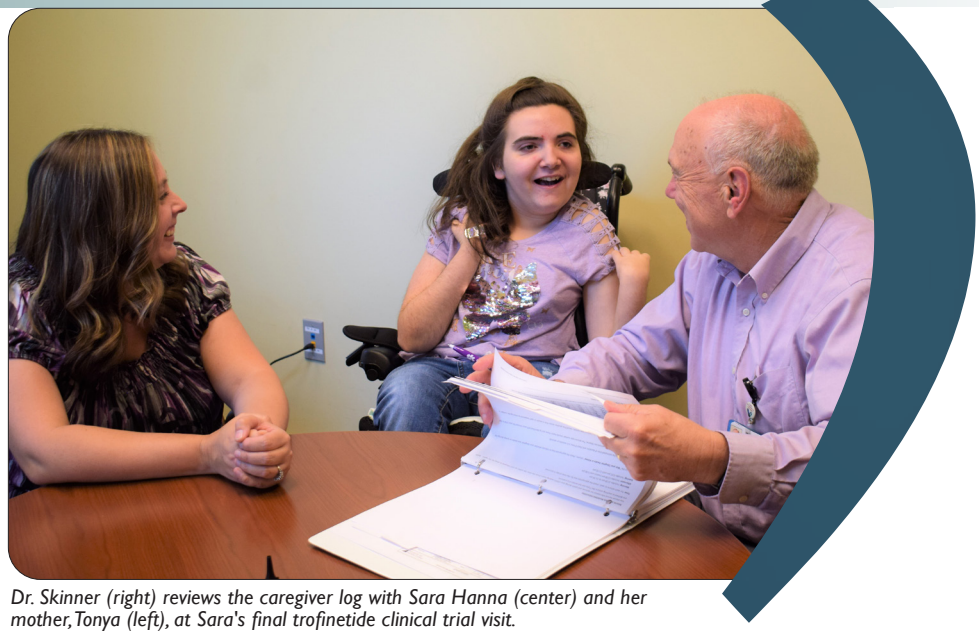
This medication was initially developed from work by the US Department of Defense as they were studying therapies for soldiers with traumatic brain injuries. Trofinetide was useful

in repairing some of the damaged brain connections and reducing inflammation. Skinner said that work led to the possibility that it may also be beneficial for patients with congenital neurological disorders like Rett syndrome. In studies of a mouse model of Rett syndrome, the drug was able to reverse many of the symptoms as well as extend the life of affected mice.

DAYBUE became available to patients in the US on April 17.

Part of the clinical trial process includes the daunting possibility of an audit by the FDA during their evaluation of the drug. GGC was the first site, and one of three in the clinical trial, to be selected for an audit. "FDA personnel visited GGC last year and combed through every detail of the trial," said Annese. "And thanks to our wonderful clinical trials team, we passed the audit with flying colors!"

"After decades of clinical research and drug development, we now have something to offer these patients and families that has the potential to improve many of the symptoms of Rett syndrome," said Skinner.



*Dr. Skinner (right) reviews the caregiver log with Sara Hanna (center) and her mother, Tonya (left), at Sara's final trofinetide clinical trial visit.*

GGC has been active in the study and care of patients with Rett syndrome for over two decades, including the National Institutes of Health-sponsored natural history study, the current Rett syndrome registry, and several prior clinical trial projects. The Center also holds a Rett syndrome clinic at Shriners Hospital for Children in Greenville.

"The approval of DAYBUE represents a long-awaited development for the treatment of Rett syndrome," said Annese. "We are excited and hopeful for the benefits this will have for our families and are grateful for those who joined the clinical trial and helped bring this drug to market."

*Cover photo: Sara Hanna surrounded by GGC Rett clinical team members, Georgia Miller, Fran Annese, Dr. Steve Skinner, and Jennifer Claphan*

# Carroll Campbell Alzheimer's Initiative

GGC is expected to receive funding in the upcoming South Carolina state budget for the Carroll A. Campbell, Jr. Alzheimer's Initiative. SC Governor Henry McMaster has requested the funding which is named in honor of former SC Governor Carroll Campbell who passed away with Alzheimer's disease in 2005. Alzheimer's disease is the most common cause of dementia and impacts more than six million Americans.

The proposed project is a collaboration between GGC; MitoSense, a research and development biotechnology company; and the US Veteran's Administration to assess a potential therapy for Alzheimer's disease aimed at improving energy production in brain cells.

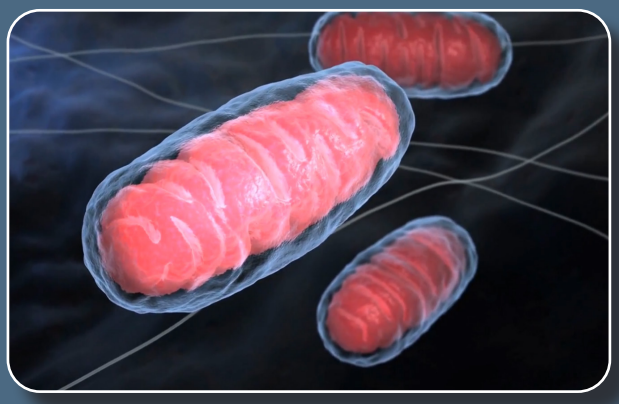


*Illustration of mitochondria (Image courtesy of MitoSense)*

The brain requires very high amounts of energy for normal activity. This energy is produced by the mitochondria, the powerhouses of the cell. Patients with Alzheimer's disease show dysfunction of the mitochondria, specifically in their brain cells, which leads to reduced energy production and is believed to be related to degeneration of those brain cells and disease progression.

MitoSense has developed a technology to replenish mitochondria through Mitochondria Organelle Transplantation (MOT™). The processes places healthy mitochondria into cells with depleted or ineffective mitochondria. The goal is to improve the energy production within the cells leading to better cellular function, reduced symptoms, and slower progression of the disease.

"This is GGC's first significant involvement in Alzheimer's disease research," said Rich Steet, PhD, GGC's Director of Research who is working on the proposed project. "We are excited about the potential of this new technology and anticipate that it could be a significant advancement for patients with Alzheimer's disease, and might also benefit our pediatric patients who have rare disorders caused by mitochondrial dysfunction."

# Recognizing Potential

## GGC post doc accepted to prestigious program

Courtney Matheny, PhD, has always had a passion for helping people. She knew from the age of 12, after learning that a friend had a congenital heart defect, that she wanted to make her mark in the world through medicine.

Matheny was originally considering medical school when she went to work in the lab of Drs. Rich and Heather Steet at the University of Georgia. "It didn't take long for me to absolutely fall in love with using zebrafish to study human disease," said Matheny, "and within six months of working for the Steets, I decided to apply to graduate school."

Upon completing her PhD at Emory University, Matheny realized that throughout graduate school she had lost some of her passion because her research wasn't directly helping anyone.

It was then that she reached out to Dr. Heather Flanagan-Steet, who had since moved her zebrafish lab to the Greenwood Genetic Center. "Even though I didn't work for her anymore, she was still my mentor," said Matheny. "She told me about what they were doing at GGC and how I could be a part of it. The compassion I have for others is what truly drives me forward in my scientific career, and rejoining the Steets' lab at GGC just felt like the right thing for me to do."

Dr. Matheny joined GGC in 2022 as a postdoctoral associate, or post doc, in the Center's Research Division.

*Dr. Courtney Matheny works with zebrafish embryos in GGC's Allin Aquaculture Facility as Dr. Heather Flanagan-Steet looks on.*

A postdoc is a position that many scientists pursue after graduate school to provide additional research experience, skills, and training to further prepare them for a faculty-level career in academia or research.

Dr. Matheny's current projects involve working with zebrafish in GGC's Allin Aquaculture Facility to better understand the causes of neurological symptoms experienced by patients with rare metabolic disorders such as congenital disorders of glycosylation and lysosomal storage disorders.

*Dr. Courtney Matheny in the Allin Aquaculture Facility*

"I love working with model organisms like the zebrafish because they are such an elegant solution to understanding our own biology and diseases," said Matheny. "Our goal should be to understand how a mutation affects the whole body and you can only do that by studying the whole organism."

Matheny's ongoing postgraduate education will expand this summer. She has been accepted to participate in the prestigious 'Zebrafish Development and Genetics' course at the Marine Biological Laboratory in Woods Hole, Massachusetts. Matheny was one of only 22 individuals selected for the course out of hundreds of applicants.

Thorough a rigorous schedule of lectures, bench experiments, and hands-on learning, Matheny hopes to gain a better understanding of the zebrafish central nervous system and new techniques that she can apply to her work at GGC.

"We are extremely excited for Courtney, as this is a tremendous educational and career development opportunity for her," said Dr. Flanagan-Steet, Director of Functional Studies at GGC. "It will also bring additional visibility to the Center in the zebrafish community - with several world-renowned scientific leaders serving on the course faculty."

Dr. Matheny hopes this experience will enhance her skills and provide her with the confidence and expertise to some day run her own research lab. "It feels like I'm at the beginning of a very exciting career and am becoming a true expert in my field of study."

Of her mentors, the Steets, Courtney said, "Anyone who works with them can tell how passionate they are and how incredibly hard they work to find answers for patients. It's what I hope someone says about me someday."

# Epigenetics

## Novel Test Reaches Milestone


Four years ago, GGC's Greenwood Diagnostic Labs partnered with London Health Science Centre (LHSC) in Ontario to launch EpiSign, a new type of genetic test. In January of 2023, GGC hit a milestone of completing 1,000 EpiSign tests.

EpiSign looks for chemical tags on a patient's DNA to identify patterns or 'epigenetic signatures' that are unique to certain genetic conditions. These chemical tags turn genes on and off at the appropriate times. If this process is altered, it can lead to abnormal expression of otherwise typical genes, which can cause disease.

Dr. Bekim Sadikovich of LHSC initially developed this testing, and he reached out in 2016 to introduce this new technology to GGC where the technology and interpretation was refined.

From those early partnerships, three labs, GGC, LHSC, and Amsterdam University Medical Centers, launched EpiSign in 2019. Greenwood Diagnostic Labs became the first lab in the United States to offer EpiSign clinically, and remains the only US lab providing this testing.

"When a patient has an epigenetic abnormality, the genes involved are usually normal in their sequence, they just are not being expressed properly," said Matt Tedder, PhD, Staff Scientist in GGC's Molecular Lab, who handles EpiSign interpretations. "This test can identify epigenomic abnormalities that other testing isn't able to find, allowing us to make a diagnosis that cannot be identified with any other test."

When it was first launched, EpiSign could identify 19 conditions. But as the field of epigenomics has advanced, scientists have identified new signatures for more conditions allowing the test to expand each year. The latest version, EpiSign v4, was launched in February and includes testing for over 70 genetic conditions – providing patients with answers that were previously not detectable by traditional methods of genetic testing.

EpiSign can also be used to help determine if a novel genetic variant identified by genome sequencing or chromosomal microarray is actually disease-causing by determining if the variant actually impacts the function of the gene.

Over the past four years, EpiSign has provided a diagnosis for many families for whom traditional genetic testing had failed to provide an answer.

"We are proud to be on the cutting edge of novel genetic testing technologies," added Tedder.

*Photo: GGC faculty and staff involved in EpiSign testing celebrate the 1,000th test.*

# GENE/GIVING WEEK

This fall, GGC will be celebrating another Gene Week, also to be known as Giving Week.

Save the dates for the following events and watch the GGC website and social media pages for details on how you can get involved.

Proceeds from all events and fundraising efforts will benefit the GGC Foundation's 'GGC Cares Fund' which provides financial assistance for GGC families who are in need of genetic consultations, testing, and/or treatment but are uninsured or underinsured. To date, the fund has provided nearly $55,000 to assist families who would not otherwise be able to afford these life-changing services

## 'Keys for Care'

Dueling Pianos Fundraiser
Thursday, September 28
Sundance Gallery in Greenwood

Enjoy cocktails, dinner, and wonderful musical entertainment!

## 13th annual Race the Helix - Greenwood

5K race and 1 mile run/walk
9:00 am Saturday, October 7
GGC's Greenwood campus

## 'Science on Tap'

GGC's Division of Education is partnering with the Clemson Center for Human Genetics for this community lecture series highlighting scientific pursuits across the Greenwood community. Details on the fall event are to be announced.

If you or your organization would like to sponsor one of the above events, please reach out to the GGC Foundation at (864) 388-1801.

*Matt and Melissa Emery with a picture of their son, Dylan*

# 'DYLAN'S LAW' REALIZED

## South Carolina Begins Krabbe Disease Screening with GGC Support

In 2019, SC Governor Henry McMaster visited GGC to sign Dylan's Law, legislation that would add Krabbe disease to the state's newborn screening test, also known as the heel prick test.

The law was named in memory of Dylan Emery, a GGC patient from Ninety Six, SC, who passed away from Krabbe disease at 11 months of age. His family worked with state legislators to add the rare disorder to the SC newborn screening panel allowing future affected infants to receive an early diagnosis with the hope of life-saving treatment.

SC began screening for Krabbe disease on May 15, adding it to the panel of 55 other genetic disorders, becoming just the 11th state to test all newborns for this rare condition.

"We have been waiting for this day," said Dylan's parents, Matt and Melissa Emery. "This is proof that Dylan's struggle and suffering will make a difference in other babies' lives."

"Because newborn screening is not a diagnostic test, all positive screens from the state must be confirmed to distinguish truly affected infants from false positive cases," said Francyne Kubaski, PhD, a staff scientist in GGC's Biochemical Lab. "That's where GGC comes in."
Kubaski and her colleagues have been validating the testing of the biomarker psychosine to help with that process.

*"We have been waiting for this day. This is proof that Dylan's struggle and suffering will make a difference in other babies' lives."*

*-Matt and Melissa Emery*

Patients with Krabbe disease are unable to make an enzyme that breaks down psychosine. The buildup of psychosine in the central nervous system leads to the loss of myelin, a protective coating on the nerves, which causes significant neurological impairment.

Patients with Krabbe are typically asymptomatic at birth, but the disease can progress quickly. The infantile form begins with feeding difficulties, muscle weakness/stiffness, and fevers. As the disease progresses, patients may experience seizures, hearing and vision loss, nerve pain, and the inability to swallow and move, with death often occurring within two years.

Treatment for Krabbe disease includes bone marrow or stem cell transplantation which allows the patient to make more of the deficient enzyme, slowing progression of the disease. Once symptoms develop, it's too late to reverse the damage that has already occurred which is why proponents have advocated adding Krabbe to the newborn screening panel.

Initial screening for all infants born in SC is performed at the state lab through dried blood spots taken from a heel stick. If the state lab identifies a low activity of the Krabbe enzyme, GGC will begin the confirmation process using these dried blood spots to test psychosine levels. By validating the testing using dried blood spots, this confirmatory testing can be performed quickly without obtaining a new sample.

"It is important to include psychosine testing to confirm the diagnosis, as some individuals will display a low enzyme level in lab testing, but in actuality they are producing a sufficient amount enzyme," said Kubaski. "This is called a pseudodeficiency, and does not cause disease."

But if enzyme levels are low and psychosine is elevated, that is a true positive result. GGC will then follow up with DNA sequencing of the gene responsible for Krabbe disease and

patients are referred for consideration of a stem cell or bone marrow transplant.

"For babies identified to have significantly elevated psychosine levels consistent with the infantile form of Krabbe disease, the time from diagnosis to treatment will be critical," said Neena Champaigne, MD, Clinical Associate Professor and Division Chief of Pediatric Genetics at the Medical University of South Carolina. "The best outcome for survival requires they undergo stem cell transplant before 30 days of life. Coordination of care between the health department, primary care provider, specialists, the GGC laboratory and families will be key."

Psychosine levels can also be measured on affected infants during treatment to determine if the transplanted stem cells are producing sufficient enzyme to decrease the toxic psychosine.

The Emery family has found comfort in knowing that Dylan's legacy lives on. "We have always feared Dylan would be forgotten," they shared. "We feel honored to be the parents of such a brave and amazing soul and are grateful to everyone who loved Dylan and helped put 'Dylan's Law' into motion."

*Francyne Kubaski, PhD, staff scientist in GGC's Biochemical Genetics Laboratory, has led the project to validate psychosine as a biomarker for Krabbe disease.*

"It is our hope that lives will be saved and other babies in South Carolina will have a better chance at survival if they are unfortunately diagnosed with this terrible disease," added the Emerys.

# Race the Helix-Upstate

## 9th annual event brings together GGC families and supporters

Runners, walkers, and GGC friends - new and old, met at Lake Conestee Nature Park in Greenville, SC for the 9th annual Race the Helix-Upstate benefitting the GGC Foundation.

The beautiful weather and a great cause brought out 190 registered participants plus dozens more supporters and volunteers to raise money for the GGC Cares Fund that provides financial support for genetic services, testing, and treatment for patients who are uninsured or under-insured.

"This event is always such a joy to be a part of," said Cady Nell Keener, Executive Director of the GGC Foundation. "It's incredibly gratifying to be able to meet so many of the families we serve and hear stories of how GGC has made such an impact in their lives. We are blessed to be able to do what we do every day."

The Shenal family, who started Race the Helix in Greenwood in 2011 in gratitude for the care their family has received at GGC, were also on hand.

*Levi Puskas, 6, a GGC patient, gives his mom, Dianna, a high five as she crosses the finish line.*

"My heart was happy seeing all the families that came out, and meeting other parents of children with genetic conditions," said Jodi Shenal, mom to Ryleigh, 12, "It was a great day!"

Race the Helix-Upstate was made possible by many volunteers and through sponsorship from numerous individuals and businesses. Special thanks to Presenting Sponsors: Bionano, Countybank, Greenwood Capital, Mutual of America, Dr. Soheil and Azita Shams, and Frank and Cathy Witney.

*SAVE THE DATE for Race the Helix-Greenwood - Saturday, October 7!*

*Stephen Shenal, center, celebrates his age group win with his family and Race the Helix-Upstate spotlight family, the Connors. L-R (Charlie, Brooks, and Wes Connor, Stephen, Jodi, Ryleigh, and Tyler Shenal.*

# EATON SUPPORTS LAB TECHNOLOGY


Eaton Corporation presented $5,000 to the GGC Foundation for the acquisition of a FlexSTAR instrument which allows for fully automated DNA extraction and isolation.

"The isolation of DNA from a patient sample is the first step for most DNA-based testing in our lab, including whole genome sequencing," said Mike Friez, PhD, Director of GGC's Diagnostic Laboratories. "By automating this process with the FlexSTAR instrument, we will be able to isolate DNA from more patient samples at once, helping us get to that critical diagnosis more quickly."

"Eaton Corporation is proud to be part of the Greenwood community, and we are committed to bringing tangible benefits to the places in which we work and live," said AJ LaGroon, Senior Human Resources Generalist for Eaton Corporation in Greenwood (pictured far right). "Technology advancement, whether through Eaton's work in energy and electrical solutions or GGC's progress in medical diagnostics, is vital to ensure the best quality of life for our communities."

"We are grateful to Eaton for this investment in the health and well-being of the patients and families we serve," said Cady Nell Keener, Executive Director of the GGC Foundation (pictured far left). "By keeping pace with the latest technological advances in genetic medicine, we are improving the quality of life for patients and families who reach out to GGC when they need that important diagnosis."

# GGC FOUNDATION CELEBRATES DONORS

GGC Foundation Board member, Tara Smith, and her husband, Brandon, opened their home for a reception honoring members of the GGC Foundation's Double Helix Society which recognizes donors who have contributed $1,000 or more over the past year. Guests were treated to wine and hors d'oeuvres, along with musical entertainment by Jarrett Smith.

GGC Director, Steve Skinner, MD, welcomed guests and shared the impact of philanthropic support on the past successes, present work, and future plans of the Center. The featured speaker was Josh Albon, 16, a junior at Emerald High School in Greenwood. Josh (left) has autism spectrum disorder and has been followed by GGC since the age of two.

"Being a person with autism has created unique opportunities and challenges for me," shared Josh. "I've been blessed to receive excellent therapy which has given me what I need to communicate with family, friends, and others and to share my unique ideas."

To learn more about the Double Helix Society and donor opportunities, please visit ggc.org/foundation

# LIGHTS, CAMERA, ACTION...

Thanks to a grant from the Fullerton Foundation, GGC is developing a studio to create a variety of video projects to support patient, student, and professional education, as well as to promote awareness of the work of the Center.

The grant is allowing for the renovation of a space in the Genetic Education Center, as well as for the purchase of equipment and the addition of a part-time videographer.

Anna Arnett has joined GGC as videographer and has been instrumental in the project's development. Her first project was a promotional video for Race the Helix-Upstate with additional videos in progress to highlight careers in genetics for middle and high school students, as well as patient testimonials to celebrate GGC's upcoming 50th anniversary in 2024.

*Arnett setting up a shot for an interview.*

The grant project was developed by the GGC Foundation and the Division of Education. Leta Tribble, PhD, Director of Education, is excited for the potential of the project. "High-quality videos are the best way to for us to engage the students we work with and get them excited about the field of genetics. We are also planning to record educational videos for other healthcare providers to expand their access to genetics content."

The project will also extend to support other divisions at GGC with the use of patient-friendly content to explain genetic testing options, promotion of diagnostic lab offerings, and sharing of patient and donor stories to grow GGC awareness and philanthropy.

# GGC Welcomes New Board Members

The GGC Foundation welcomed four new members to its Board of Trustees in January.

GGC Foundation trustees serve as mission ambassadors by cultivating, securing, and stewarding philanthropic funds that advance the Center's mission for the benefit of the patients and families served by GGC. They also manage all fiduciary responsibilities and activities of the Foundation.

Helen Campbell of Florence, SC owns Organization Solutions and is the mother of a son who has benefited from a genetic diagnosis through the Greenwood Genetic Center.

Ted Pitts of Lexington, SC is the President and CEO of Wilson Kibler Commercial Real Estate. He has also served in the US military, the SC House of Representatives, as Deputy Chief of Staff and Chief of Staff in the SC Governor's office, and as President and CEO of the South Carolina Chamber of Commerce.

Arthur Radcliffe of Greenwood, SC is a Regional Sales Director for Vidya Herbs. A Navy veteran, he has served on various boards, including the Lander University Alumni Board, Natural Products Association Foundation, and Main Street United Methodist Church Foundation.

Kay Self of Greenwood, SC has been involved in the community by volunteering at dozens of organizations. She is also a Board member for SCBio and Executive Director of Vision Greenwood.

"We welcome these four new Foundation trustees," said Ray Wilson, PhD, chair of the board. "Each of these new trustees brings enthusiasm and unique expertise to the Center's mission which will allow us to support the GGC and all of the patients, families, and students they serve."

GGC also announces the appointment of two new members to its Holdings Board of Directors in April.

The eleven-member GGC Holdings Board was created in 2022 as part of the GGC/MUSC membership agreement. This body is responsible for the selection of the six GGC-appointed members of the Center's Board of Directors along with exercising other defined powers under the joint agreement.

Christen Davis of Ninety Six, SC is Senior Director of Quality at Lonza. She is also a member of the GGC Foundation Board of Trustees.

Sam Konduros, JD, of Simpsonville, SC is the Founder and President of SK Strategies, a consulting firm specializing in economic development, strategic planning, and innovation. Konduros is the former President and CEO of SCBio.

"I'm so proud that this extraordinary patient-focused, life sciences R&D hub is located in my home state of South Carolina, and I am extremely honored to serve on the GGC Holdings Board to help advance its bold and compelling mission," said Konduros.

"We are so pleased to welcome Christen and Sam to the GGC Holdings Board," said Chris Przirembel, PhD, chair of the Holdings Board. "Both of these individuals are longtime supporters and champions for our mission and bring talents that will help the Center grow and prosper as a leader in the field of medical genetics."

Top row: Campbell, Pitts, and Radcliffe; Bottom row: Self, Davis, and Konduros

## EMPLOYEE UPDATES

Congratulations to the following GGC employees who have excelled in their roles and have earned achievements and/or promotions in recent months...

**Katy Drazba, MS, CGC** of GGC's Columbia clinic was promoted to Lead Genetic Counselor. She has been serving as interim in that role since last summer.

**Grace Hollingsworth, MS, CGC**, of GGC's Greenwood clinic passed her board examination and is now certified by the American Board of Genetic Counseling.

**Dan Moats,** a technologist in GGC's DNA lab, passed his American Society for Clinical Pathology certification exam for credentialing medical laboratory professionals and was promoted to Laboratory Technologist II.

**Mattie Piotrowski** of GGC's Greenwood clinic has been promoted to Lead Genetic Assistant.

**Mia Smith**, a technologist in GGC's biochemical lab, was promoted to Laboratory Technologist II.

The Greenwood Genetic Center is a nonprofit institute organized to provide clinical genetic services, diagnostic laboratory testing, educational programs and materials, and research in the field of medical genetics.

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