Greenwood Genetic Center

Summer
2021

A Newsletter for the Friends of the Center

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GGC's SC Birth Defects Prevention Program Recommends More Folic Acid

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More Folic Acid than You Think!

GGC makes new recommendations for women of childbearing age

The journey to becoming parents did not start as an easy one for Chris and Alicia Jones of Conway, SC.

"We suffered the heartache of miscarriage and the unimaginable pain of burying our firstborn child," recalls Alicia.

In 2007, Joseph "Hamilton" Jones was born at just over one pound and 12 inches. He had spina bifida, hydrocephalus, and a heart defect. Hamilton passed away just 2 hours and 40 minutes later.

"My husband and I were left choosing a small white casket, not much larger than the size of a shoebox for him to be buried in," said Alicia. "It tore our hearts out leaving the hospital without him."

Spina bifida, along with anencephaly and encephalocele, are classified as neural tube defects (NTDs), anomalies caused by the failure of the neural tube to close very early in development. In the late 1980s, it was discovered that folic acid, when taken as a supplement by women prior to and early in pregnancy, could reduce the risk for these life-altering, and sometimes life-threatening birth defects. Since that time, the SC Birth Defects Prevention Program at the Greenwood Genetic Center has made it their mission to educate all women of childbearing age on the importance of taking folic acid.

"Over the past three decades we have had much success in reducing the incidence of NTDs in South Carolina, a state that once had the highest incidence of these birth defects in the US," said Jane Dean, RN, Statewide Coordinator of the Program. "These educational efforts, along with a surveillance program and folic acid fortification in cereal grain flours have led to a 60% reduction in NTDs in SC."

That translates to 70 healthy babies born each year without an NTD.

Through educational campaigns including billboards, brochures, social media, and attendance at health and bridal fairs, the program has encouraged all women of childbearing age to take 0.4 mg of folic acid each day. Women who have had a prior pregnancy affected with an NTD, like Alicia, are instructed to take even more folic acid, 4 mg/day, to prevent a recurrence.

"While we have been pleased with the declining rate of NTDs in SC, we still continue to see approximately 25 of these potentially preventable birth defects in our state each year," said Roger Stevenson, MD, who oversees the prevention program. "Failing to consider additional preventive measures is simply not acceptable."

Dean and Stevenson's efforts to monitor NTDs in SC, which were most recently published in the *Journal of Pediatrics* (226:186, 2020), led to the conclusion that for women in a high-risk state like SC, folic acid fortification in flour and grains does not provide sufficient protection against many NTDs. Dean and Stevenson suggest that the rate of NTDs could be further reduced by increasing the folic acid dosage to 4mg per day, not only for those with a prior affected pregnancy but for all women of childbearing age.

"We shouldn't wait until a family has suffered a tragic loss or a child is born with lifelong disabilities if we can prevent the first occurrence through a higher dose of folic acid," said Dean.

Higher doses of folic acid, a water-soluble B vitamin, have been shown to be generally safe. Potential side effects of high doses are uncommon, but some such as diarrhea, skin rashes, and sleep disturbances have been reported in certain populations, along with the potential to mask vitamin B-12 deficiency and lowering the effectiveness of anticonvulsant medications.

Thus, the SC Birth Defects Prevention Program proposes that the standard preventive dose of folic acid for all women of childbearing age be increased to 4 mg/day during the three months prior to conception and the first month after conception.

Currently, women on a typical diet receive approximately 100-150 micrograms/day of folic acid through fortification of cereal grain flours. Therefore, the recommendation to use 4 mg/day of folic acid remains the same whether or not the woman uses folic acid fortified food products.

Chris and Alicia Jones with Brynleigh, Ellieana, and Preston

The additional supplementation should be prescribed as folic acid alone rather than by increasing the number of multivitamins, as the dosage of other vitamins may become excessive.

Women who experience any potential side effects of high doses of folic acid should discontinue the 4 mg/day dose and return to a multivitamin with 0.4 mg or a prenatal vitamin with 0.8 mg.

"Following the loss of Hamilton, I enrolled in GGC's NTD research program and began taking a folic acid supplement, increasing my dosage to 4 mg/day when we began planning to expand our family," said Alicia. "We are now the proud parents of three healthy children, Brynleigh, Preston, and Ellieana. We are thankful for the research of the Greenwood Genetic Center and the efforts spent towards supporting the families affected by genetic defects and preventing them from occurring in the future."

For more information, contact Dean at jane@ggc.org or 1-800-676-6332.

Billboards touting the new folic acid recommendation have been placed across SC.

SKINNER RECOGNIZED BY ALMA MATER

GGC Director, Steve Skinner, MD, was awarded an honorary Doctor of Science degree from Wofford College at their spring 2021 commencement exercises. Skinner graduated from Wofford in 1975 prior to enrolling in medical school. He joined GGC as a pediatrician and medical genetics resident in 1987 and became director in 2011.

"Steve has a gentle manner and genuine care for each patient he encounters and has been a driving force in GGC's commitment to treating genetic disorders," said GGC cofounder, Roger Stevenson, MD. " Since assuming the role of director, he has worked tirelessly to expand GGC's impact, both medically and economically through the development of treatment initiatives and partnerships."

"The honorary degree is the highest form of recognition offered by the college," said Nayef H. Samhat, President of Wofford College. "We are indeed grateful to Dr. Skinner for his important and scholarly work throughout his distinguished career that has brought reflected honor to his alma mater."

Photo credit: Wofford College

ZIATS HONORED BY ACMG FOUNDATION

GGC medical genetics resident, Catherine A. Ziats, MD, was presented with the David L. Rimoin Inspiring Excellence Award by the American College of Medical Genetics and Genomics (ACMG) Foundation at the virtual ACMG annual meeting in April. The award recognizes trainees or junior faculty for their outstanding platform presentation.

During the ACMG conference, Ziats presented her work in identifying a gene within a family that appeared to cause a more severe response to COVID-19. Her talk was entitled "Alterations in respiratory epithelial gene *SPDEF* segregate with severe disease in a family with variable response to COVID-19 infection."

"I am very honored to have received this prestigious award and for the recognition by the ACMG," said Ziats. "I am also extremely grateful to the family members who agreed to participate in this study, who despite their difficult time found it important to attempt to advance our knowledge on host genetic factors associated with severe COVID-19 infection,"

Ziats donated her award money to the GGC Foundation to promote similar ongoing research.

BUCH COMPLETES MAPP FELLOWSHIP

Physician assistant, Laura Buch, MSPAS, PA-C, completed a Metabolic Advanced Practice Provider (MAPP) fellowship at GGC in April. The two-year program, sponsored by Sanofi Genzyme, involved hands-on clinical genetics training with a focus on the diagnosis and medical management of individuals with lysosomal storage disorders (LSDs). Buch was GGC's first MAPP fellow and will remain with GGC's Greenville office serving both general genetics patients, as well as individuals with LSDs and other metabolic disorders.

"Laura completed her Metabolic Advanced Practice Provider fellowship with dedication and motivation," said Curtis Rogers, MD, Senior Clinical Geneticist and Buch's mentor for the MAPP fellowship. "She did an amazing job, and we are blessed and excited that she will continue working at the Greenwood Genetic Center."

"Over the past two years of my MAPP fellowship, I have had the privilege to work closely with our LSD patient population, both through clinical care and research opportunities. I also gained a better understanding of the behind-the-scenes efforts of our laboratories who bring diagnostic results to families," said Buch. "I look forward to continuing my career at GGC working with this outstanding group of families, colleagues, and mentors."

'COBRE' GRANT

NIH funds first 'Center of Biomedical Research Excellence' in human genetics


GGC's Flanagan-Steet will lead one of the COBRE research cores through her pioneering use of zebrafish as a model organism.

Genetics defines an individual's unique characteristics that – coupled with lifestyle habits and other environmental factors – determine susceptibility to cancers, hypertension, high cholesterol, arthritis, diabetes, Alzheimer's disease and numerous other ailments.

The National Institutes of Health (NIH) has tasked Clemson University and the Greenwood Genetic Center with unlocking these genetic codes through a new $10.6 million grant to establish the Center of Biomedical Research Excellence (COBRE) in Human Genetics in collaboration.

The award funds an initial five-year phase of the COBRE, which can continue for 15 years, positioning the Clemson-GGC collaboration as a global leader in the scientific advancement of human genetics. The NIH COBRE program provides a long-term investment in the advancement of medical research around a central theme. This is NIH's first COBRE specifically focused on human genetics.

Trudy Mackay, PhD, Self Family Endowed Chair in Human Genetics and Director of the Clemson Center for Human Genetics based in Greenwood, will lead the COBRE in Human Genetics along with GGC's Director of Research, Richard Steet, PhD, and Robert Anholt, PhD, Clemson's Provost's Distinguished Professor of Genetics and Biochemistry.

> ***"Imagine having the opportunity to immediately sequence the genome of a baby and being able to quickly identify their susceptibility to disease. We can't change their genes, but we can change their environment, significantly improving preventative care. If we know the susceptibility, we can make non-pharmaceutical interventions to improve many people's quality of life. And of course, knowing the underlying biology can help guide pharmaceutical intervention as well."***
>
> ***- Trudy Mackay, PhD***

GGC has collaborated closely with the Clemson University Center for Human Genetics since the facility opened in Self Regional Hall on the Greenwood campus in 2018.

"Merging the expertise of Clemson's genome science with the patient-driven focus of the Greenwood Genetic Center is very powerful," Steet said. "The theme of this COBRE is comprehensive – covering common disorders like cardiovascular disease, cancer, neurodegenerative diseases, as well as very rare genetic disorders. We take a lot of pride in that breadth, as it gives our collaborations and the efforts of this COBRE room to grow."

"GGC is honored to be part of this first-ever NIH COBRE in the field of human genetics," said Steve Skinner, MD, GGC Director. "By combining the Greenwood Genetic Center's 47 years of expertise in providing quality medical genetics services with the research talent and computational power of the Clemson Center for Human Genetics, patients and families impacted by both common and rare genetic diagnoses will reap the benefits."

"This grant truly raises the profile of both Clemson University and the Greenwood Genetic Center, and I am proud that our collaboration has the potential to make a difference for so many people. It is powerful to think of how many lives might be saved by learning more about the genetics behind some of these devastating diseases," said Clemson University President Jim Clements.

Cover photo (L-R) Anholt, Mackay, and Steet will lead the NIH's first COBRE in Human Genetics.
Photo credit: Clemson University

COBRE funding is intended to support professional development and research goals of junior faculty, allowing them to grow their laboratories and develop programs to compete for their own NIH grants. Four projects are funded in the initial COBRE cycle:

Andrei Alexandrov, PhD, Assistant Professor of Genetics and Biochemistry at Clemson's Center for Human Genetics, will be analyzing human nuclear long non-coding RNAs to identify potential targets for new treatments for cancer and viral diseases.

Miriam Konkel, MD, Assistant Professor of Genetics and Biochemistry at Clemson University, will study how and why transposable elements, or "jumping genes," can move around the human genome and alter genetic expression. This movement may contribute to neurodegenerative diseases like Alzheimer's.

Heather Flanagan-Steet, PhD, Director of Functional Studies at GGC, will study genetic mutations that can cause neurological and cognitive impairment, skeletal abnormalities and even early infant death. She pioneered the use of zebrafish to model rare inherited diseases to investigate gene function and disease pathogenesis.

Fabio Morgante, PhD, Assistant Professor of Genetics and Biochemistry at Clemson's Center for Human Genetics, will analyze genetic data from 500,000 people to develop models that can predict cardiovascular disease. His models will take into account ancestry, ethnicity and environmental factors that can affect disease susceptibility.

GGC Discovery Provides Hope

Research identifies mechanism and possible treatment for movement disorder

Balance issues came first, then hand and eye tremors, seizures, and learning delays. Chloe Murphy, 15, pictured right, and her parents had no answers until GGC's Dr. Mike Lyons suggested whole exome sequencing to analyze all 20,000 genes.

Chloe's test results identified a variant in a gene called *NUS1*. "But, it was a variant that no one had seen before, so we weren't sure that it was the answer," said Lyons. "We reached out to the research team at GGC to help clarify its meaning."

GGC researchers ran experiments on skin cells from Chloe and were also able to find two other patients with *NUS1* variants and similar symptoms. They also developed zebrafish models that mimicked the *NUS1* variants seen in these patients. Zebrafish and humans share 70% of the same DNA, making them a powerful animal model to better understand genetic disorders.

These studies showed several interesting findings. First, these *NUS1* variants seemed to cause excess cholesterol accumulation within cells. Second, zebrafish with the *NUS1* variants displayed abnormal swimming behaviors, akin to Chloe's movement disorder. The researchers also saw improvements in the zebrafish when treatments targeting cholesterol accumulation were applied.

"These studies not only confirmed that this variant was indeed the cause of Chloe's symptoms, providing a long-awaited answer for her family, but they also gave us the opportunity to better understand this rare diagnosis and consider potential treatments," said Rich Steet, PhD, Director of Research at GGC.

The Murphy family has found hope through their research participation.

"Our journey so far has been to find the 'why.' We felt like once we could determine this, we could better understand what could help Chloe," said her mother, Jessica. "Our hope now is that these breakthroughs with *NUS1* will lead to a solution to improve her health."

Read more of Chloe's story at The Gene Scene at ***www.ggc.org/the-gene-scene***.

Rett Center of Excellence

GGC earns re-designation by International Rett Syndrome Foundation

Every day that GGC has a patient with Rett syndrome on the schedule, you'll find the clinic staff decked out in purple, the official color of Rett Syndrome awareness.

GGC became closely intertwined with the Rett syndrome community over 15 years ago with the start of the NIH Rett syndrome natural history study. GGC's 'Rett Team' enrolled patients at GGC as well as through travel clinics across the US with a goal of better understanding the clinical features and genetics of this rare disorder

In 2016, GGC was among the first group of institutions recognized as a Rett Syndrome Clinical and Research Center of Excellence. That designation was renewed in 2021 by the International Rett Syndrome Foundation (IRSF). GGC joins other distinguished organizations such as Boston Children's Hospital, Children's Hospital of Philadelphia, and Vanderbilt University with this designation.

The award recognizes GGC's continued dedication to providing best-in-class clinical care for Rett syndrome, a rare neurological and developmental disorder. It comes with funding support from IRSF and membership in IRSF's Center of Excellence Network.

To qualify as a Center of Excellence, clinics must be led by a physician director with expertise in Rett syndrome, provide health care services that meet the unique needs of individuals with Rett, be involved in clinical trials, and contribute to a clinical registry.

"We are honored to continue this prestigious designation as a Rett Syndrome Center of Excellence," said Steve Skinner, MD, GGC's Director who also leads the Center's Rett syndrome program. "We have had the privilege of working closely with IRSF and many families impacted by Rett syndrome, and we renew our commitment to serving them with compassionate care and the latest in medical breakthroughs."

Clinical Care

GGC currently leads a multidisciplinary Rett syndrome clinic at Shriners Hospital for Children in Greenville, SC that serves families from across the southeastern US. This bimonthly clinic provides comprehensive services including clinical genetics, genetic counseling, neurology, developmental pediatrics, and physical and occupational therapy in a single visit.

Clinical Trials

GGC is also currently involved in two clinical trials to investigate treatments for Rett syndrome.

Acadia Pharmaceuticals is sponsoring a trial of trofinetide, a compound that has shown great promise in early studies in alleviating many symptoms of Rett. GGC had originally planned to enroll six patients but currently follows seven participants. The trofinetide trial is expected to continue until enough data has been collected to submit the drug for FDA approval.

GGC is also a clinical trial site for Epidiolex, an oral CBD derivative that already has FDA approval to treat seizures in other rare disorders. GGC was the first US site to begin enrolling patients in this study sponsored by GW Pharmaceuticals. In addition to seizure control, this drug is also being investigated for other common features of Rett including breath-holding, involuntary hand movements, and anxiety.

For Fran Annese, GGC's Greenwood clinic manager and clinical research study coordinator, her work with Rett syndrome is a passion. "I have been honored to be a small part of the journey for these families over the past 15 years. To see the amazing growth in our understanding of this condition that now holds the promise of treatment is so exciting."

Clinical Research

After enrolling over 1200 patients, the NIH Rett syndrome natural history study will end in July. The study has generated a great deal of information about this rare disease and has led to advances in treatment and medical management.

Skinner has been a principal investigator on the natural history study and currently serves on the medical advisory board to develop a new protocol to continue to gather data through a new Rett syndrome disease registry.

Biobanking

In addition to clinical research, GGC serves as the biobanking site for the entire Rett research community. GGC collects and receives patient samples from across the US. These samples can be used for testing by GGC labs and are also available to other researchers for their studies.

"We've made tremendous progress in the study of Rett syndrome, but we have much more work to do," said Skinner. "With the collaborative efforts of IRSF and the other Centers of Excellence around the world, we are confident that better treatments, and even a cure, are on the horizon."

What is Rett Syndrome?

Rett syndrome is a rare neurodevelopmental disorder that affects 1 in 10,000 females. Typically caused by a mutation in the *MECP2* gene, affected individuals begin to experience delays and developmental regression at 6-18 months. Other features include loss of speech and purposeful use of hands, involuntary hand movements, loss of mobility and muscle tone, seizures, scoliosis, breathing issues, and sleep disturbances.

Top: Elizabeth Wilkerson enjoys the park following her trofinetide study visit.
Bottom: Mackenzie Noecker (front, center) poses with her GGC Rett team after a clinical trial visit.

Inspiring the Workforce

The outreach programs of GGC's Division of Education have two goals - to improve genetic literacy and encourage career exploration among the students they serve.

The first goal is measured with each student who comes to campus or is visited by the Gene Machine or Helix Express Before and after the activity, brief assessments are completed to evaluate their knowledge of genetics. In the 11 years of the program, 100% of classes have gained genetics knowledge during their GGC experience. Regardless of the students' intentions to enter a genetics-related career, this knowledge will make all students better, more informed healthcare consumers.

The second goal of encouraging students to pursue careers in genetics is tougher to measure; however, GGC is starting to see the fruits of those efforts within its own workforce.

Mattie Sullivan *(pictured above with the Gene Machine)* joined GGC in April as a genetic assistant in the Greenwood office. She supports the clinical team by obtaining insurance authorizations, taking family histories, and assisting as needed during patient visits. She is also helping to implement GGC's new eConsults and eVisits to improve patient communication and access.

Sullivan, who aspires to become a genetic counselor, participated in visits from the Gene Machine twice while a student at D.W. Daniel High School in Central, SC.

"I appreciated getting to delve deeper into genetics - learning about the different career paths and actually getting to pipette and practice with my own DNA," she said. "I didn't get to hear much about genetic counseling through my regular high school classes, so having GGC come speak and show it on the screen was always so cool to me."

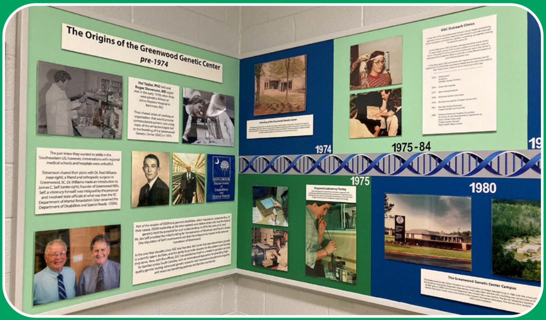
Caleb Hinzman has plans to attend graduate school to become a physician assistant. In the meantime, he's joined GGC as a technologist in the cytogenetics laboratory running diagnostic tests and providing answers for patients and their families.

Hinzman attended Greenwood Christian School and visited GGC's Genetic Education Center with his biology class.

"I learned how rapidly the knowledge of DNA was developing and how relatively new a lot of the science was," he said. "That visit helped reinforce my decision to pursue a career in biology even though I did not know exactly what I wanted to do, yet."

"It is so gratifying to meet new GGC employees and learn that they were once students of our outreach programs," said Leta Tribble, PhD, GGC's Director of Education. "Our patients are currently benefitting from their early exposures to genetics, and that knowledge will stay with them as they pursue their future career paths in their chosen medical field."

"As a high school student I never would have imagined I would have a job at GGC pre-grad school," said Sullivan. "But now I get to look outside my window every day, see the Gene Machine, and be reminded of my journey so far."



Education Center opens GGC Museum

Students and guests visiting GGC's Genetic Education Center can now take a peek back in time to learn about the history of genetics and the Greenwood Genetic Center.

Last year, as the pandemic brought GGC's outreach educational efforts to a halt, the education team turned their attention to projects that they had not previously had time to work on - one of those was creating a GGC museum.

The result is a hands-on exhibit space showing the progression of genetic technology through the years.

The GGC museum includes exhibits on cytogenetic, molecular, and biochemical lab testing with examples of the equipment used through the years including early gel electrophoresis equipment and the Center's first next-generation DNA sequencer.

The museum space also includes a timeline mural of the important events in GGC history, from the story behind the founding of the Center through present day. The timeline also highlights many of the scientific discoveries made at the Center.

"When students visit the Genetic Education Center, we share state-of-the-art technologies and the latest information about genetic disorders," said Leta Tribble, GGC's Director of Education. "Now, they will not only leave with an appreciation of where we are in this exciting field but also how we got to this point, with a focus on how GGC has contributed to the advances in genetic medicine."

Above: The timeline mural in GGC's new museum highlights the founding of the Center in 1974 and the Center's growth and contributions to the field throughout the past 47 years.

BAKE SALES leading to BREAKTHROUGHS

Family-led organizations are key to advancing rare disease research

> "The support, guidance, and shared experiences of parents who have been on this journey before us has been invaluable, and has helped us get through some of the most difficult times of our lives."

Rare diagnoses can leave families feeling a host of emotions including confusion, fear, and worry. When your child has been given a diagnosis that you've never heard of before, feelings of isolation often become overwhelming.

Now, in the days of greater global connection through the internet and social media, there are more and more family-led organizations and foundations connecting families with rare diseases across the country and around the world. These organizations, most often started by families who searched and found no resources when they were diagnosed, provide a lifeline for the newly diagnosed - support from those who have been there, information when it's hard to find, ways to become engaged in a community, and hope for the future through research.

Andrea Miller was a nonprofit administrator and lawyer when her daughter, Bianca, now 11, was diagnosed with a rare condition called PMM2-CDG at 15 months.

CDGs, or congenital disorders of glycosylation, are a large and growing group of rare inherited disorders. There are over 100 known genes that cause the various CDGs. The clinical features vary but often include developmental delay, low muscle tone, skeletal defects, and seizures.


Andrea Miller with her daughter, Bianca, hiking in support of the Angel Bandit 5K on CDG Awareness Day in 2019

In 2014, Miller founded CDG CARE, a volunteer-based 501(c)3 nonprofit to provide support, resources, and education to families newly diagnosed with CDGs.

"The support, guidance, and shared experiences of parents who have been on this journey before us has been invaluable, and has helped us get through some of the most difficult times of our lives," said Miller. "While the benefits we receive from this patient network are ongoing and transition to different issues as Bianca gets older, the ability to be able to give back and help parents who are just beginning their CDG journey has been equally, if not more, therapeutic and rewarding for our family."

GGC researchers have recently received funding from CDG CARE and another family-led organization, the Yash Gandhi Foundation, to support transformative research into rare disease treatments.

GGC's Director of Functional Studies, Heather Flanagan-Steet, PhD, has been awarded a $50,000 grant from the Frontiers in CDG Consortium and CDG CARE. Dr. Flanagan-Steet and her team will use zebrafish models of CDG to investigate the mechanism of a new drug, epalrestat, to treat PMM2, one of the more common types of CDG.

"Like many rare disease advocacy organizations, CDG CARE initially focused its initiatives on directly assisting patients and families," said Miller. "By 2018, our efforts began to expand into

Philanthropy Supports Lab Modernization

The GGC Foundation has received funding from The Self Family Foundation, as well as from private donors, to support an upgrade to a new Laboratory Information Management System (LIMS) to improve modernization efforts for GGC's diagnostic laboratory and research divisions.

LIMS is a powerful new software technology used to manage diagnostic workflow from the time a test is ordered until a final clinical report is delivered. Updating GGC's LIMS will add precision, speed, capacity, and efficiencies to the Center's diagnostic and research programs.

The Self Family Foundation provided $250,000 to the GGC Foundation for the new LIMS with an additional $200,000 contributed from private donors.

"We are grateful for this investment from The Self Family Foundation and many steadfast individual donors that will allow GGC to keep pace with the explosion of genetic testing and ensure that all patients receive accurate and timely results when they need them most," said GGC Director, Steve Skinner, MD. "This powerful LIMS will improve the turnaround time and accuracy of the genetic testing our patients depend on."

THE
SELF FAMILY FOUNDATION
Legacy · Community · Opportunity

funding scientific research to advance the diagnosis and treatment of CDG."

The organization has a few committed donors and applies for grants and industry funding; however, 60-70% of funds are raised though individual family-led events such as t-shirts sales, 5K fun runs, charity auctions, wine tastings, and social media giving campaigns. Many of these events are held in conjunction with CDG Awareness Day which is celebrated each year on May 16th.

"These grants from family-led organizations are very special to us," said Flanagan-Steet. "These families work so hard to raise the money for these projects, and they are placing their hope and trust in our work. It is both an honor and a tremendous responsibility, one that the entire team involved in the work takes very seriously."

Through the support of these fundraisers, relationships with industry partners, and the volunteer efforts of the organization's Medical Advisory Board members, CDG CARE has successfully funded over $200,000 in research projects from basic science to clinical research.

The Yash Gandhi Foundation has also awarded a $90,000 grant to GGC to continue current work into the investigation of over 60 small molecules which have the potential to treat I-cell disease, also known as mucolipidosis II (MLII).

GGC has a long history of involvement with MLII and has received funding from the Yash Gandhi Foundation for several years.

Families gathered at the February 2020 Rare Disease Day Symposium and CDG CARE Family Conference in San Diego, CA.

The Foundation was started by the family of Yash Gandhi who died of I-cell disease at age eight. The family-led organization hosts fundraising events including an annual 'i Run for i-cell' 5K race.

Kavi Gandhi, Yash's younger brother, has taken an active role in the Foundation and even spent the summer of 2020 as an intern in GGC's research lab learning more about the science behind this rare disease.

"I always knew that the support our Foundation gave to researchers was important and powerful, but I had no concrete picture of what that meant," said Gandhi. "Getting to spend time in the GGC labs and witnessing with my own eyes the important and revolutionary work that is being done has been indescribably gratifying."

"Working so closely with CDG CARE, the Yash Gandhi Foundation, and other family-led organizations gives such meaning to our work," said Flanagan-Steet. "Our team is bolstered by seeing the direct impact we have on the patients and families who look to us for answers and hope."

"By collaborating, patient advocacy groups and researchers can make faster progress toward new treatment options and better patient outcomes, which can improve the lives of all persons and families affected by rare diseases such as CDG," shared Miller.

GGC's EpiSign Clinically Validated

The Greenwood Genetic Center, working with collaborators at Lawson Health Research Institute in Canada and the University of Amsterdam, published a study in February issue of *Genetics In Medicine* that provides clinical validation of EpiSign, a molecular genomics test that diagnoses rare, heritable neurodevelopmental conditions. GGC has been the sole US laboratory provider of this novel diagnostic test since 2019.

EpiSign analyzes changes that affect gene expression rather than the gene sequence. Researchers have found that certain genetic disorders display unique patterns, or epigenetic signatures, allowing for a diagnosis through EpiSign when traditional genetic testing has been uninformative. The current study analyzed data from early EpiSign testing to validate the ability of the novel test to make a diagnosis. Epigenetic signatures have been identified for over 40 genetic disorders.

EpiSign is the only test in the world that has been clinically validated for epigenetic testing for these types of genetic disorders, and in the US, is only available through GGC.

"Patients with rare diseases often wait years and undergo numerous exams and tests before receiving a correct diagnosis, if one is found at all," says Matthew Tedder, PhD, a staff scientist at the Greenwood Genetic Center who was involved in the study. "EpiSign provides an additional high-yield diagnostic tool for clinicians to include in their evaluation of patients with undiagnosed diseases, providing better medical management for patients and hope for their families."

Right: Tedder performs a step in the EpiSign assay to analyze changes in gene expression

A VIRTUAL SUCCESS

GGC's 7th annual Race the Helix - Upstate was held virtually on May 8 with participation from as far away as San Diego, CA, Great Britain, and Paris!

"Last year's Upstate race had to be cancelled at the last minute as the pandemic had just taken hold," said Brooke Smith, MS, CGC, a genetic counselor in GGC's Greenville office and Race the Helix-Upstate committee chair "With the months of planning that goes into these events, we decided last winter to stay as a virtual race to ensure the safety of our friends and supporters."

However, with vaccine availability and declining COVID-19 numbers, many participants were able to gather with colleagues, friends, and family to complete their walk or run in support of the GGC Foundation and the patients and families served by GGC. Bionano Genomics, one of the event sponsors, encouraged many of their employees around the globe to participate and held their own race event. (below, center). GGC's Greenville office employees planned a fun outing to run and walk together as well (below right).

"While we are all looking forward to hosting an in-person event next year, we will also continue to keep a virtual component as well," said Smith. "With GGC's global presence and influence, we're so excited that anyone can participate from wherever they are."

We are grateful to the many sponsors who made Race the Helix-Upstate possible:

Presenting Sponsor - Mutual of America
Helix Gold Sponsors - Bionano Genomics, Alka Chaubey and Ridhi, First Citizens Bank, Frank and Cathy Witney
Education Booth Sponsors - Ascend Cares Foundation, A Friend of GGC in honor of all affected by autism, Greenwood Capital, Industrial Compressor Solutions, Luftmann Contractors
Walk the Helix Sponsors - Lonza, SC Tel-Com
Helix Silver Sponsors - Davis Marketing, Raymond James, Regions Bank
Shenal Family Sponsors - David and Sarah Everman, The Manning Law Firm, PA, New York Butcher Shoppe, Curtis and Debbie Rogers, Sadler Hughes Apothecary, Team Charlie Connor

GENETICS ON THE AIRWAYS

DNA TODAY
HOSTED BY KIRA DINEEN

Looking for your next podcast? GGC has recently been featured on two popular podcast series.

Ray Louie, PhD, Assistant Director of GGC's Molecular Diagnostic Lab and Kellie Walden, MS,CGC, Director of Diagnostic Development, were interviewed for 'DNA Today,' an award winning podcast and radio show exploring how genetics impacts health. They discussed epigenetics and GGC's EpiSign test which has recently been clinically validated to diagnose over 40 rare genetic disorders *(see p.9)*. 'DNA Today' was the 2020 People's Choice award winner for best science and medicine podcast. Listen at dnapodcast.com

'Taking the Pulse,' a weekly podcast that covers the healthcare sector in the Carolinas, featured GGC Director, Dr. Steve Skinner in April. Skinner shared the story of GGC's founding, explained the Center's clinical, diagnostic, research, and educational mission, and discussed his take on the future of genetics in medicine. 'Taking the Pulse' is produced and hosted by Nexsen Pruet Health Care Practice Group. Listen at thepulsehealthcast.com/

TAKING THE PULSE
A Health Care Podcast

In Loving Memory

Karen Buchanan (1958-2021)

Karen Buchanan started working at GGC on February 14, 1995 - Valentine's Day. Looking back, that seems an appropriate milestone considering how much she loved working at the Greenwood Genetic Center.

For 26 years, Karen, who passed away in January from complications of COVID-19, served as Dr. Roger Stevenson's trusted assistant.

“Karen was deeply involved in all aspects of the Greenwood Genetic Center's evolution," recalls Stevenson. "She was involved in preparing our first large NIH grant, planning for the growth of the campus as we built the JC Self Research Institute and the Treatment Center, and supporting the research efforts of gene discovery and birth defects prevention."

Cindy Skinner, RN joined GGC as Dr. Stevenson's nurse on the same day as Karen recalling her colleague and friend as a 'computer whiz.'

"She helped me tremendously with my graphs and charts, and even my grammar," said Skinner. “Karen loved her work at GGC. She loved the scientific aspect of it, and being involved in the publications and genetic discoveries."

"Karen brought devotion and her extraordinary talent to benefit both GGC, and me personally, throughout these 26 years," added Stevenson, "But Karen was more than that. She was an integral part of the Center’s brain thrust."

Beyond GGC, Karen was a devoted wife, mother, and new grandmother, and was very active in her community. She enjoyed volunteering at the Bowers Rodgers Children's Home reading stories to the children. Karen served in Cub Scouts and Boy Scouts for 15 years leading Cub Scout Pack 270, working with the Blue Ridge Council's Cub resident camp, and serving as Scoutmaster of Troop 313. She was very proud that her sons, Patrick and Eric, both achieved the rank of Eagle Scout.

Always an avid learner, Karen spent much of her free time reading, researching, writing, and gardening.

"Karen will be missed, but she will always be a part of GGC,” said Skinner.

Career Milestones

Recognizing employee achievements

While this past year has been challenging in many ways, GGC is pleased to recognize several employees who have excelled in their work and have achieved recognitions and advancement.

Lori Bassett, MS, CGC, was promoted to Director of Communications.

Melea Jefferson was promoted to Team Lead of GGC's Greenville office.

Talitha Kay was promoted to Assistant Human Resources Manager.

Andrew Lay was promoted to Bioinformatics Specialist in GGC's Diagnostic Laboratories.

Savanna Lyda advanced to Laboratory Technologist Level II in GGC's Molecular Diagnostic Laboratory.

Sydney Mann advanced to Laboratory Technologist Level II in GGC's Molecular Diagnostic Laboratory.

Hannah Moore, MS, CGC, was promoted to Lead Clinical Genetic Counselor.

Laura Pollard, PhD, was promoted to Lead Director of GGC's Biochemical Genetics Laboratory.

Ellen Richardson, MS, CGC, passed the board examination to become certified by the American Board of Genetic Counseling.

Falan Talbert was promoted to Accession Specialist in GGC's Diagnostic Laboratories.

Falecia Thomas, MS, CGC, passed the board exam to become certified by the American Board of Genetic Counseling.

Meg Wilkes, MS, CGC, passed the board exam to become certified by the American Board of Genetic Counseling.

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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