Greenwood Genetic Center

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A Newsletter for the Friends of the Center

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New Technologies Leading to More Diagnoses

The Greenwood Genetic Center has recently acquired two new state-of-the art instruments to enhance genetic testing capabilities.

SAPHYR

GGC is the first organization in South Carolina to acquire Bionano's Saphyr instrument for advanced genetic testing capabilities.

The Saphyr uses a novel technique called optical genome mapping (OGM) to identify structural variations in the genome at a higher resolution and in a more cost-effective and timely manner than traditional cytogenetic testing.

"Optical genome mapping combines the capabilities of three clinically available tests, karyotyping, microarray, and fluorescence in-situ hybridization, and has 1000X higher resolution than traditional chromosome analysis," said Barb DuPont, PhD, Senior Director of GGC's Cytogenetics Laboratory. "We can effectively replace three tests with this single assay that is faster, less expensive, and will also identify genomic changes that we would never have been able to detect with any other test."

Nikhil Sahajpal, PhD, Laboratory Genetics and Genomics fellow at GGC, is leading the Center's OGM project under a grant from the American College of Medical Genetics and Genomics Foundation.

Sahajpal and DuPont with GGC's Saphyr instrument Inset: (courtesy of Augusta University): a Bionano chip with DNA imaged at single molecule resolution using optical genome mapping.

"Optical genome mapping allows us to analyze long pieces of DNA and reconstruct the chromosomes, similar to putting together a puzzle," said Sahajpal. "OGM overcomes some of the limitations of other methodologies and allows us to identify structural genetic changes that have been significantly understudied because of the lack of technology."

Molecular lab technologists (L-R) Kevin Thomas, Khirston Howard, Seth Shearin, and Kevin Babson during training on the NovaSeq X Plus.

Sahajpal gained experience with OGM during his post-doctoral fellowship at Augusta University and is applying the new technology to several GGC patient cohorts where a genetic diagnosis has been lacking. Current studies include patients with neural tube defects, limb anomalies, and prenatal cases where there are ultrasound anomalies, but genetic testing has so far been uninformative.

"Early testing has already been able to identify a causative genetic variation in a prenatal case where current standard-of-care tests had fallen short," added Sahajpal. "We are driven by the hope of providing a diagnosis that couldn't have been found any other way and ending what can often be a very long diagnostic odyssey for patients with rare diseases."

GGC envisions that OGM may prove to be a significant advancement in genetic diagnostics.

"GGC has recently launched a precision medicine initiative (p.4)," said GGC Director, Steve Skinner, MD. "Part of that initiative aims to improve our ability to analyze the genome and provide answers to patients who remain undiagnosed. OGM is another tool in our arsenal, and we are excited by the potential for OGM to provide an accurate and rapid diagnosis for patients who have these difficult-to-detect variants."

NovaSeq X Plus

GGC's Genomic Sequencing Program received a boost in October with the acquisition of the NovaSeq X Plus instrument. This top-of-the-line DNA sequencer by Illumina was purchased with funds from the Center's affiliation with MUSC.

The NovaSeq X Plus joins GGC's NovaSeq 6000 instrument to expand the capacity for whole genome sequencing which allows for an analysis of the entire genome. The testing identifies mutations throughout the patient's entire DNA sequence or areas where a patient may have extra or missing genetic material that causes their symptoms.

"This new instrument enables increased sample throughput compared to our current NovaSeq 6000," shared Jennifer Lee, PhD, Lead Director of GGC's Molecular Diagnostic Laboratory. "It incorporates improvements in the chemistry that allow for higher accuracy, performance, and speed - ultimately leading us to make patient diagnoses more quickly."

The new instrument also has shorter sample run times and a lower cost per sample for whole genome sequencing.

"The addition of both the NovaSeq X Plus and the Saphyr instruments are improving our ability to identify diagnoses for patients that are not detectable by traditional testing methods," said Mike Friez, PhD, Director of GGC's Diagnostic Laboratories. "As part of GGC's long-standing commitment to patient care, and with the new precision medicine initiative (see p. 4), we are making significant advances in diagnostic capabilities, with the ultimate goal of guiding management and improving the quality of life for the patients and families we serve."

RUDY EARNS SUPERVISORY AWARD

Natasha Rudy, MS, CGC, a genetic counselor in GGC's Greenville office was presented with the 2023 Victoria A. Vincent Genetic Counselor Supervision Award by the genetic counseling program at the University of South Carolina School of Medicine.

Natasha was nominated by the graduating genetic counseling class because she exhibits the qualities of an ideal supervisor: providing balanced feedback, cultivating the critical thinking process, building confidence, allowing autonomy with respect for student opinion, and fostering a collegial relationship.

"Natasha was selected because of her ability to establish a supervisory relationship through which students are both supported and challenged, to provide constructive feedback that promotes student growth, to provide students with excellent learning opportunities while maintaining high quality patient care, and for serving as an example of what a genetic counselor should be," said outgoing program director, Janice Edwards, MS, CGC.

Natasha was also recognized along with outstanding supervisors from other Master's programs at the National Society of Genetic Counselors annual meeting in Chicago in October.

SCHOLARSHIP HONORS RETIRED GENETICIST

Curtis Rogers, MD, served GGC's patients and families for 40 years as a clinical geneticist. Upon his retirement last year, he was honored by the organization's human resources (HR) department with the creation of a scholarship in his name. GGC's Director of HR, Janet Still, and HR Manager, Talitha Kay, funded the scholarship which covers the cost for two students from Connie Maxwell Children's Ministries in Greenwood to attend the Center's Junior Genetic Scholars summer camp for high school students.

Cameron Turnnidge, a 9th grader, and Dana Baca Espinal, a 12th grader, (pictured left with Rogers) attended this year's camp thanks to this scholarship. GGC has offered the popular summer camp since 2018 to provide engaging hands-on genetic activities to high school students who are interested in genetics.

"Dr. Rogers has passion, both for his patients and for sharing his love of genetics with the many students who have had to good fortune to train with him," said Still. "Knowing this, Talitha and I wanted to honor him by providing educational opportunities for students to encourage their interest in genetics and to see firsthand how science provides answers and hope."

GGC HOSTS FIRST MIDDLE SCHOOL CAMP

Since 2018, GGC's Junior Genetics Scholars Camp for high school students has been a popular summertime activity for older teens interested in genetics. In the summer of 2023, the Center's Division of Education extended this opportunity to middle school students.

The first middle school genetics camp was held in June at GGC and hosted 15 rising 6^{th}-8^{th} graders. Activities focused on basic biotechnology skills with labs on DNA isolation, forensic analysis, and calico cat genetics interspersed with presentations by GGC educators, DNA model building, Jeopardy-style games, and genetics-themed bingo. "It felt like I was a real detective and scientist," shared one participant at the end of the week.

"We've seen such enthusiasm and excitement as we've expanded our school year programs to serve more middle school students," said Ashley LaVerdure, Operations Manager for GGC's Outreach Education Program (pictured right with two middle school campers). "By engaging students at the middle school level, we are opening up so many exciting career opportunities to them at a time when they are being first introduced to genetics. We look forward to this becoming a regular summer offering for these younger students."

Straight A's

GGC's Genomic Discovery Program Supports Precision Medicine Initiative

Providing compassionate care starts with a patient's first connection with GGC with the ultimate goals of delivering an accurate diagnosis and personalized treatments.

GGC's new Precision Medicine Initiative (PMI) is a framework of projects to enhance all aspects of the patient's experience with genetics. By working to improve the four A's - Access, Analysis, Answers, and Action - GGC aims to support families throughout their experience with genetics and ultimately provide the right treatment for the right patient at the right time.

ACCESS

The Access component of GGC's PMI was developed to ensure that anyone in South Carolina in need of genetics services would be able to receive them.

Quality genetics services begin before the patient ever comes into contact with GGC. By streamlining the referral process, reducing wait times, and recruiting additional clinical personnel, GGC will be better able to handle the growing demand for these services. Additional projects to improve access are being implemented through the expansion of eVisits (see p.5) and the greater use of technology to offer genetics consultations and genetics education for non-genetics providers to hasten the ordering of genetic testing.

ANALYSIS

As genetic and genomic technology expands, so does the ability to make diagnoses that were once unattainable.

Through the Analysis component of the PMI, GGC is exploring new technologies such as RNA sequencing and optical genome mapping (see p.2) to generate data that may provide answers for patients when more traditional testing methods have been uninformative.

"The GDP has the potential to directly impact so many families who have yet to find the answer and the treatments they've been looking for."

-Mike Lyons, MD

Director of Clinical Services and Lead of GDP Workgroup

ANSWERS

The crux of the Answers component of the PMI lies in the development of GGC's Genomic Discovery Program (GDP).

The GDP is a cross-divisional program designed to identify timely answers for undiagnosed patients and specific treatments to improve the quality of life for patients and their families.

The GDP is a new resource for clinicians to access after clinical genomic testing methods have not provided a clear answer. It involves a collaborative effort of clinicians, diagnostic laboratory faculty, and research scientists to provide enhanced care for families by offering access to novel genomic technologies and analytic methods, shortening the time to diagnosis for patients with rare diseases, and identifying personalized treatments specific to the patient's medical needs.

The GDP working group has developed three tracks:

1. **Discovery**
   When a patient's genetic testing results have not been able to identify a cause, the team will review these undiagnosed cases to determine the next best approaches and technologies to employ to find the answer.

2. **Resolution**
   When a patient's prior test results identify uncertain variants that may or may not be causative, the GDP team will review the results to determine what types of experiments could be conducted.

3. **Treatment**
   Once a diagnosis is confirmed, the GDP will attempt to connect patients with existing therapies and explore novel treatment options.

The GDP workgroup has been meeting regularly to develop a database of eligible patients and define the workflows and criteria for inclusion into this program. Patients must be referred through their GGC provider for consideration. The GDP plans to begin the review of the first group of patients in January.

"We are very excited to begin enrolling patients in our new GDP program, and believe that this has the potential

to directly impact so many families who have yet to find the answer and the treatments they've been looking for," said Mike Lyons, MD, Director of Clinical Services and Lead of the GDP Workgroup. "This team that combines expertise from GGC's clinics, diagnostic labs, and research divisions is committed to ensuring access to a diagnosis and treatment for children and adults with suspected or known genetic conditions."

Meg Keating, MS, CGC, Mike Lyons, MD, and Aubrey Rose, MS, CGC, three clinical members of the GDP review inclusion criteria for the program.

ACTION

With the ultimate goal of providing treatments and improving symptoms and quality of life, the Action piece of the PMI will expand clinical trials both at GGC and through collaborations with colleagues at MUSC.

GGC's expansion into researching neurodegenerative disorders such as Alzheimer's disease through the Carroll Campbell Alzheimer's initiative (see p.9) also holds the promise of treatments for this common condition as well as applications of this technology to other rare mitochondrial or metabolic disorders.

GGC's PMI, while existing as a standalone program of the Center, is also being incorporated into a broader collaborative precision health initiative with MUSC. This precision health initiative expands upon GGC's access, analysis, answers, and action pillars to create a Precision Health Institute to house cross-cutting clinical, research, and educational initiatives.

"GGC's Precision Medicine Initiative, combined with the broader precision health collaboration with MUSC, holds an incredible potential for us to use information about a person's genetic material, its interaction with the environment, and their lifestyle to prevent, diagnose, and treat disease more efficiently and effectively," added Steve Skinner, MD, Director of GGC. "This is groundbreaking work that will benefit patients and their families for generations to come."

Grant Expands Access for Infants and Toddlers

GGC has been awarded a grant of $191,779 from the SC Center for Rural and Primary Care. The two-year project, 'Expansion of eVisits to ensure statewide access to genetics care for BabyNet-eligible infants and toddlers', aims to improve access to care, shorten wait times, and provide earlier genetic diagnoses for this vulnerable group of patients.

BabyNet is South Carolina's early intervention program for infants and toddlers under three years of age with developmental delays or who have conditions associated with developmental delays. Through GGC's relationship with the SC Department of Disabilities and Special Needs, all BabyNet-eligible infants and toddlers are offered a genetics evaluation. This new funding will expand GGC's eVisit program offering these asynchronous consultations to all BabyNet-eligible families.

"eVisits remove several barriers to care such as travel, childcare, and missing work, and they also allow patients to be evaluated and start genetic testing much more quickly – within a week of referral compared to a weeks-long wait for an in-person appointment," said Mike Lyons, MD, Director of Clinical Services at GGC and the principal investigator on this project.

The project will also facilitate the referral process for early interventionists in rural areas of South Carolina and provide educational materials for providers and families on the benefits of an early genetic evaluation.

"Genetic evaluations for infants and toddlers enrolled in BabyNet offer the possibility of identifying the underlying cause for these developmental delays at an early age, allowing for more targeted therapies, management, and support for families," said Lyons. "eVisits show great promise in helping us provide the same high-quality patient care in a more timely and convenient manner."

"We are proud to support GGC's expansion to provide rural services," added Andrea Mitchell, Program Manager for the SC Center for Rural and Primary Healthcare. "Proactive and innovative programs such as this are critical to ensuring that quality care is available and accessible to improve rural health in our state."

GGC leads identification of new cause for genetic kidney disease

At age 11, Aaron Ritz woke up with side pain. His pediatrician sent him to the hospital where he was found to be in complete renal failure. He started dialysis the next day.

"Aaron was diagnosed with polycystic kidney disease, which he likely had from birth, but aside from being small for his age, we had no signs that anything was wrong," said Aaron's mother Brandy Ritz. "In less than 24 hours, our world was turned completely upside down."

There are two forms of polycystic kidney disease (PKD). The autosomal dominant form affects approximately 1/1000 individuals. It typically presents in adulthood and is most often caused by a single genetic mutation in one of two genes, *PKD1* or *PKD2*. The autosomal recessive form of PKD is less common (1/20,000) and presents prenatally or in early childhood with two mutations within the same gene.

Aaron's nephrologist suggested genetic testing due to the unusual nature of his clinical presentation, so he was seen at the Greenwood Genetic Center. Testing revealed no mutations in the genes most commonly associated with polycystic kidney disease, but he did have a single variant in a gene called *NEK8*.

"*NEK8* is known to be a rare cause of autosomal recessive PKD when there are two mutations present, and typically there are also other extra-renal manifestations such as liver and pancreatic involvement," said Rich Steet, PhD, GGC's Director of Research. "But Aaron, who had no other organ involvement, only had a single genetic change in *NEK8* which had never before been associated with kidney disease."

Given the puzzling results, Steet embarked on a research project to try and clarify this unexpected finding. By reaching out to other genetic testing laboratories and using GeneMatcher, an online tool that connects clinicians and researchers who have an interest in the same gene, Aaron's genetic evaluation grew into a major international collaboration. Steet connected with geneticists in the US, the Netherlands, the United Kingdom, Canada, Belgium, Denmark, the Czech Republic, and Romania, and ultimately identified 21 individuals within 12 families who had both polycystic kidney disease and a single variant in *NEK8*. All of these patients also had a second normal copy of the *NEK8* gene.

The Ritz family on a 2023 trip to Colorado. Inset: Aaron, 11, during his first dialysis, less than 24 hours after his initial diagnosis, August 2018

Numerous additional experiments were conducted by the collaborative group to confirm that these single variants were causative for PKD and to better understand how they cause kidney disease.

"Our experiments suggest that the genetic variants identified alter the NEK8 protein function in a very specific way," said Steet. "It appears that the abnormal protein produced by the *NEK8* variant in these families prevents certain other proteins from moving to cilia, structures in kidneys that help them function normally. When these proteins fail to localize to the cilia, severe cystic kidney disease develops."

After a rough disease course which included severe hypertension, a stroke, and the removal of both diseased kidneys, Aaron had a complicated, but ultimately successful kidney transplant at age 12 and now, at 16, is thriving.

"Aaron has a regimen of medications that he will have to take for the rest of his life, but he's doing great – working a part-time job and playing golf for his high school team," said Brandy. "It all happened so fast, we didn't really have time to process what was going on at the time, but now, to have the worst behind us and to have a clear diagnosis means everything!"

Steet noted that this is important work that expands our understanding of both the *NEK8* gene and polycystic kidney disease. "In the future, clinicians should pay attention to single *NEK8* variants in patients with PKD, include this gene in genetic testing panels, and consider the possibility of a single *NEK8* variant as disease-causing," he added.

The collaborators published these new findings in *Kidney International*, a journal of the International Society of Nephrology, in August.

COBRE Project Leads to Prestigious R01 Grant

In 2021, the Clemson University Center for Human Genetics and GGC received a 5-year, $10.6M grant from the National Institutes of Health (NIH) to establish the first Center of Biomedical Research Excellence (COBRE) in Human Genetics.

Research in the COBRE in Human Genetics focuses on understanding the mechanisms by which genetic variation affects both rare and common diseases. The COBRE in Human Genetics supports four research projects from investigators that tackle several of the challenges facing human genetics.

GGC's Associate Director of Research, Heather Flanagan-Steet, PhD, was one of the four researchers tasked with leading a COBRE project. She and her team have been studying genetic mutations that can cause neurological and cognitive impairment, skeletal abnormalities, and even early infant death. Their work on rare diseases largely involves the development of zebrafish models to investigate the gene function and the process by which mutations in these genes cause disease.

A major goal of the COBRE is to develop a critical mass of investigators to improve infrastructure in the organizations' area of research who are able to compete effectively for independent research funding.

Flanagan-Steet's project recently graduated from the COBRE system and the project has been awarded a highly competitive R01 grant from the NIH.

The new, two-year, $500,000 funding will support this research team's continued focus on congenital disorders of glycosylation (CDGs). CDGs are a group of rare inherited diseases caused by mutations in genes involved in the attachment of sugar molecules to proteins and lipids - a process known as glycosylation. Errant glycosylation affects the function of cells in every part of the body.

There are many types of CDGs, and symptoms vary from mild to severe, but often include low muscle tone, poor growth, developmental delay, liver disease, stroke-like episodes, and heart problems.

Under this new R01 funding, GGC's research team is using zebrafish models of the most common of these CDG disorders, PMM2-CDG, to understand how altered glycosylation causes disease symptoms with the idea that this information will point to new therapies for patients.

"Zebrafish are a very useful model system for studying CDGs because they allow us to examine early development in transparent embryos," said Allen Wu, a Clemson graduate student working on the project. "We are hopeful that our research will provide new insights into these diseases."

Photo: GGC's researchers involved in the R01 granted project (L-R): Allen Wu, Courtney Matheny, PhD, Heather Flanagan-Steet, PhD, Lynn Rimsky, and Kali Wiggins.

EMPLOYEE EXCELLENCE

Congratulations to the following GGC employees who have excelled in their roles and have earned recognition in recent months...

Kyle Carroll of GGC's Greenville clinic was promoted to Lead Genetic Assistant.

Chandler Couick of GGC's Microarray Laboratory was promoted to Lab Techologist level II.

Ben Hilton, PhD, Assistant Director of GGC's Cytogenetics Laboratory, earned board certification from the American Board of Medical Genetics and Genomics.

Chelsi Jeter was promoted to Manager of the Allin Aquaculture Facility.

Carder Jones was promoted to Bioinformatic Analyst.

Aneta Kaczmarczyk PhD, Assistant Director of GGC's Biochemical Genetics Laboratory, earned board certification from the American Board of Medical Genetics and Genomics.

Talitha Kay was promoted to Human Resources Manager.

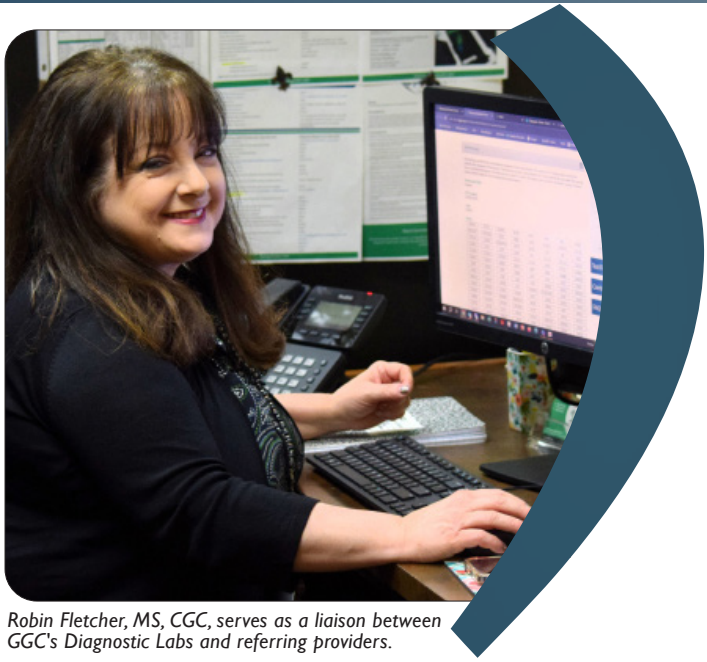
Wesley Patterson, MSPA, PA-C, PhD, a physician assistant in GGC's Greenwood clinic, was honored by the Greenwood Chamber of Commerce as an Under 40 Star.

Alison Romagnoli was promoted to Billing Operations Manager.

Kevin Thomas of GGC's DNA Diagnostic Lab was promoted to Lab Technologist, level II.

Melinda Todd was promoted to Billing Coordinator.

Kali Wiggins was promoted to Assistant Manager of the Allin Aquaculture Facility.



Robin Fletcher, MS, CGC, serves as a liaison between GGC's Diagnostic Labs and referring providers.

DIAGNOSTIC JOURNEY

Employee experiences GGC's compassionate care from the patient's perspective

Robin Fletcher, MS, CGC, left the field of genetic counseling to teach high school while trying to adopt a child. After more than seven years in the classroom, she returned to a career in genetics and joined GGC as a laboratory genetic counselor, not realizing that her new job would finally help her get the medical answer she had long sought after.

"The support I have received along with the expertise of clinicians and laboratory directors at GGC has meant more to me than I can express."

-Robin Fletcher, MS, CGC
Laboratory Genetic Counselor

"Even as a young child, I was never able to run. I couldn't catch my breath. My muscles would start burning, and for a while I missed a lot of school with frequent infections," recalls Robin. "I remember being aware of the fact that this was different from my classmates."

At age 20, she contracted mononucleosis, and from that point forward "I've been tired and it's never gone away."

In addition to fatigue, Robin's list of symptoms has grown to include intractable hypertension, chronic headaches, cardiac arrhythmia, muscle weakness, kidney disease, unexplained anemia, and hypothyroidism, to name a few. Her fatigue became so severe that as a teacher, she struggled to write on the whiteboard or hold her hair dryer while getting ready for school.

Over the years, Robin has undergone countless procedures including MRIs, CT scans, EMGs, a lumbar puncture, nerve conduction studies, and a muscle biopsy. She has visited dozens of specialists, sometimes getting the impression that they wondered if she was imagining things.

"The implication being that I was making it up, that it was all in my head," shared Robin. "That, honestly, was the most difficult part of this journey."

After returning to the field of genetics in 2014, Robin, now 56, recalled being in a meeting reviewing GGC cases when one of the clinical geneticists commented in regards to another patient that 'hypertension in your 20s is never normal.' She left that meeting and walked down the hall to see Dr. Mike Friez, the Director of GGC's Diagnostic Labs, to ask if the lab could run an exome test on her since she developed hypertension at age 28.

Robin's exome test, which identifies mutations in the coding regions of all 20,000 genes still didn't provide an answer, but it did allow her to rule out a few difficult-to-diagnose conditions.

In 2020, with the pandemic at its height, Robin, while sitting on numerous Zoom meetings, noticed a new symptom. Her right eye was noticeably drooping. Her weakness was also progressing which led her to consider that she may have an issue with her mitochondria, the part of cells that produces 90% of the energy used by the body.

"During my genetic counseling training, little was known about mitochondrial disorders, so it wasn't part of our curriculum," she said. "But as these conditions became better understood, I started thinking this might be a good fit for what I was experiencing."

In 2021, GGC's labs were adding a new analysis to look for duplications and deletions in the mitochondrial genome. Mitochondria carry circular DNA that is separate from the more familiar chromosomes and DNA in the nucleus of the cell, which is what had been studied in Robin's exome test. She asked GGC Associate Lab Director, Raymond Caylor, PhD, to run her recent muscle biopsy sample during the validation process for this new test.

Caylor added Robin's DNA sample to his very first run, and when the results were complete, he came to Robin's office, closed the door, and said 'I think we found something.'

Robin had a very large deletion of her mitochondrial genome. Three-fourths of

her mitochondrial genome was missing in a significant proportion (20-25%) of her mitochondria.

This was the answer she had been looking for! It explained her fatigue; it explained most of her symptoms, and most importantly gave her peace of mind.

"Dr. Caylor was concerned about sharing this diagnosis of a progressive disease," recalled Robin. "But my overwhelming emotion was gratitude because I finally had an answer to what had been happening."

"My boss at the time, who had helped with interpreting my test results, came by later that day and asked if I needed to go home - was I OK," she said. "And I was OK. I finally had a diagnosis after decades, essentially a lifetime, of searching."

"Finding the reason for my symptoms has meant more to me than I can express. It means I'm not lazy. I'm not imagining things. It's not in my head. It's not just nerves. I'm not a hypochondriac. It's real."

"The GGC lab did for me what they have done for so many families across South Carolina and beyond," added Robin, "They provided a resolution to my diagnostic journey."

Fletcher and Raymond Caylor, PhD, review a mitochondrial testing report.

"Although my condition is progressive, living with uncertainty was far worse for me. Knowing that what I have is real and that my symptoms have a name, has allowed me to move forward and focus on what I can do to manage my limitations," said Robin.

"One of the things that I realize as an employee of the GGC is that our lab, our researchers, our clinicians will go as far as they possibly can to try and obtain answers for families," she added. "I can say that I would not know what was wrong with me if not for the expertise of the people here.

I am eternally grateful to GGC, both for giving me a job and for giving me a diagnosis.

I work with amazing people."

Campbell Alzheimer's Research Update

GGC received $2M in funding from the state of South Carolina this year to develop the Carroll Campbell Alzheimer's Initiative in memory of the late SC governor who died of the disease in 2005. The funding is supporting a collaborative effort between GGC, MitoSense, and the Veteran's Administration to advance the study of Mitochondrial Organelle Transplantation (MOT™) as a potential therapy for Alzheimer's disease.

Normal brain activity requires very high amounts of energy which is produced by the mitochondria, the powerhouses of the cell. Patients with Alzheimer's disease often exhibit 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, a biotechnology company, has developed MOT™ to replace depleted or ineffective mitochondria with healthy ones. 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 technology will also be investigated for its potential to treat other rare mitochondrial and metabolic disorders.

The Campbell Alzheimer's Initiative will begin with the installation of a Biospherix instrument in GGC's McAlhany Family Center for Collaborative Research in Greenwood. This self-contained instrument isolates mitochondria from healthy donor cells under conditions that will allow for their use in human patients. GGC would be the only organization in South Carolina, and one of the few in the US, with the capacity to make these mitochondria. Once the Biospherix instrument is operational, GGC will generate and distribute the isolated mitochondria to their partners for pre-clinical research studies and eventually for possible clinical trials.

GGC is also recruiting two positions to contribute to the development and optimization of therapies, and the testing of the mitochondria in zebrafish disease models under this new Alzheimer's Initiative.

Photo: Rich Steet, PhD, Director of Research, is leading GGC's initiatives in Alzheimer's disease research which will be housed in the McAlhany Family Center for Collaborative Research

GGC WELCOMES NEW GENETIC COUNSELORS

Genetic counselors play a vital role in both GGC's clinics and laboratories. In the clinic, genetic counselors offer personalized risk assessment and educate patients and families about all aspects of their diagnosis while providing support and resources . Laboratory genetic counselors work directly with referring healthcare providers to answer questions, support their test ordering, and follow-up on any positive test results. GGC recently welcomed three new genetic counselors to our team.

Kenya De Leon, MS, (top) joined GGC's Columbia office providing genetic counseling in general and oncology clinics. She earned a BS in genetics from Clemson University and an MS in genetic counseling from the University of South Carolina School of Medicine. Kenya also has Spanish language fluency and is a certified medical interpreter.

Alexandra Finley, MS, CGC, (center) joined GGC's Diagnostic Labs where she serves as a liaison between the labs and their network of referring healthcare providers. Alex has experience as a prenatal genetic counselor and is interested in the multidisciplinary integration of communications, graphic design, and education within genetics to promote holistic healthcare. She earned BS degrees in biochemistry and technical writing and communication from the University of Minnesota and an MS in genetic counseling from the University of Utah.

Nisha Pandya, MS, (bottom) joined GGC's Charleston office where she provides genetic counseling in the general genetics clinic. Nisha earned a BS in biological sciences from North Carolina State University and an MS in genetic counseling from the University of South Carolina School of Medicine.

"GGC has an amazing group of nineteen genetic counselors who work in all aspects of genetic care," said Katy Drazba, MS, CGC, GGC's Lead Genetic Counselor. "We welcome Kenya, Alex, and Nisha to our team of counselors who are some of the most intelligent, compassionate, creative, and hardest-working in the profession."

EDUCATION HOSTS FIRST COUNSELING INTERN

Training the next generation of genetic counselors is an important task. Each semester, genetic counseling students spend weeks in one of GGC's clinical offices to gain valuable experience in real-life clinical settings. They practice drawing pedigrees, coordinate genetic testing, and explain very complex concepts to patients and their families under the supervision of one of GGC's exceptional board-certified genetic counselors.

UNC Greensboro's genetic counseling program requires students to spend one of their rotations in a non-clinical setting to gain exposure to the growing opportunities for genetic counselors to pursue non-traditional roles. Many students work with genetic research teams or in a genetic testing lab, but Coltrane Beck-Chance, a rising second-year student, chose education.

During his six weeks with GGC's Division of Education, Coltrane observed and assisted with hands-on STEM activities onboard the Gene Machine mobile science lab. He also developed a presentation about the ethics of genetic testing for science teachers at GGC's summer workshop and shared his journey to becoming a genetic counselor with high school students during GGC's summer camp.

"We were thrilled when Coltrane reached out and proposed this rotation," said Leta Tribble, PhD, Director of Education at GGC. "While most people think of genetic counselors in terms of their direct patient care roles, they are skillfully trained to translate complex medical information which makes them natural educators for other populations as well, including students, teachers, and other healthcare professionals."

"There are so many transferable skills between education and working in the genetics clinic," said Coltrane. "As I am presenting to a class, I am able to learn in real-time if my explanations of difficult topics are making sense. Tailoring to my audience is one of the most important things I will learn to do - to make sure that my patients are getting the most out of the genetic counseling session."

Partners in Progress

Greenwood Genetic Center
Where Compassion Inspires Progress

MUSC Health
Medical University of South Carolina

MUSC and GGC's membership agreement took effect in May of 2022. In the ensuing 18 months, both organizations have prioritized the development of relationships that are helping to build upon the initial agreement with a goal of enhancing genetics services for all citizens of South Carolina.

With several active joint working groups and the support of the highest levels of both GGC and MUSC administrations through the Joint Operating Committee, the benefits of the affiliation are tangible and growing...

- As outlined in the membership agreement, MUSC has matched $954,056 raised by the GGC Foundation.
- MUSC provided $1.3M for GGC to purchase the NovaSeq X Plus, a new state-of-the-art DNA sequencing system, to expand upon the efficiency and throughput of the Center's Genomic Sequencing Program (see p.2).
- In November, GGC representatives and MUSC leadership met with several consultants from the University of Alabama at Birmingham's precision medicine effort to discuss a framework for collaboration. The following day the 1st annual Precision Health Research symposium was held at the Storm Eye Institute on the MUSC campus and featured talks by GGC's Director of Research, Richard Steet, PhD, and Associate Director of Research, Heather Flanagan-Steet, PhD, on the Genomic Discovery Program and other precision medicine activities at GGC (see p.4). The symposium also covered topics ranging from cardiovascular genetics to precision oncology, and helped initiate ideas to expand the MUSC/GGC partnership in precision health.
- GGC's Division of Education and MUSC are co-hosting the Mobile Laboratory Coalition's Annual Meeting in July of 2024 on the MUSC campus in Charleston. The meeting will bring together educational organizations that offer mobile outreach STEM programs across the US for workshops, mobile lab tours, networking, and curricula development.
- The Education Working Group is planning a Project ECHO (Extension for Community Healthcare Outcomes) for medical genetics. Project ECHO is an innovative telementoring program designed to create virtual communities of learners by bringing together non-genetics healthcare providers and genetics experts from GGC and MUSC using brief lecture presentations and case-based learning. The GGC-MUSC education work group is conducting provider surveys to assess the needs of non-genetics providers in improving access to genetics care for their patients.
- GGC is transitioning to MUSC's Epic system which will include both the Epic electronic health record system as well as a new and improved laboratory information management system called Beaker. This transition will improve patient care by removing barrier to the flow of information between GGC's clinics and diagnostic labs. It will also include a new Genetics and Genomics module that will provide currently lacking clinical decision support when genetics variants are found. The new platform will also simplify the patient check-in process for both patients and clinics reducing manual data entry and improving efficiency.

Save the Date 2024

The Greenwood Genetic Center will be celebrating our 50th anniversary in 2024!

"Ever since Roger Stevenson, MD, and Hal Taylor, PhD, founded the Greenwood Genetic Center in 1974, GGC has been at the forefront of advances in the field of medical genetics while never compromising on providing the most compassionate care to the patients and families we serve. 2024 will be a year of celebration of this tremendous legacy and rich history along with excitement about what we can do in the next 50 years!

In 2024, the GGC Foundation will launch Care Reimagined, a campaign to support our Precision Medicine Initiative (see p. 4). With this bold plan, GGC is poised to remain a leader in genetic care for decades to come..

We hope you will make plans to celebrate with us with events throughout 2024. Watch GGC's website (GGC.org) and follow our social media accounts to keep up with all that will be happening next year!

Thank you for your continued support of our mission!"

Steve Skinner, MD
Director

50TH Anniversary
Greenwood Genetic Center
Leading the Way Since 1974

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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