Greenwood Genetic Center

Winter 2022

A Newsletter for the Friends of the Center

Rare Roses

Art exhibit displays beauty in all p.4

Photo: Jack Robert Photography

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GGC researchers uncovering how genes cause disease

GGC PA Advocates for Growth of Profession in Genetics

Wesley Patterson, PhD, MSPA, PA-C, CAQ-Peds is a passionate advocate for the physician assistant/associate (PA) profession.

Patterson recently earned a PhD in Healthcare Genetics and Genomics from Clemson University with his dissertation project, 'Genetics and Genomics Education Among Physician Assistants.' The project consisted of a literature review of PA genetics education along with surveys of practicing PAs to learn about their knowledge of and training in genetics. He also reached out to PA training program directors to gather data about their curricula and training needs in topics related to medical genetics.

And his project was not simply an academic exercise.

When he's not in clinic caring for patients or counseling families about their test results on the phone, Patterson is working on ways to educate and engage other PAs in his chosen specialty of clinical genetics.

"Wesley is one of the most enthusiastic providers that I have had the pleasure to work with," said Leta Tribble, PhD, Director of Education at GGC, and also a member of Patterson's dissertation committee. Tribble and Patterson have collaborated on a number of projects to improve and enhance genetics instruction for PA students as well as practicing PAs.

As part of his interest in improving the genetics curricula in PA training programs, Patterson is part of a five-week genetics lecture series for the University of South Carolina School of Medicine's PA students. He also provides genetics lectures for PA students at his alma mater, Jefferson College of Health Sciences, Presbyterian College, North Greenville University, and West Liberty University. In addition, Patterson offers shadowing opportunities and mentorship for students in several PA and MD/DO programs.

His work also extends to currently practicing PAs to not only encourage them to consider genetics as a specialty, but also to improve their knowledge of genetic disorders within their current area of practice.

"A knowledge of genetics and genetic disorders is not only vital for those of us practicing in genetics clinics, but it is also important across most all medical specialties, including family medicine, cardiology, and orthopedics," said Patterson. "If a PA can identify a genetic condition, or the need for a genetics referral, that will lead to a more timely and accurate diagnosis and ultimately improve the outcome and quality of life for that patient."

Dr. Wesley Patterson promotes careers in genetics for PAs. Photo by Dr. Jessica Cooley Coleman.

Patterson has shared his work at professional conferences across the US including the Clinical Genetics Advanced Practice Provider Conference, the Academy of Physician Assistants (AAPA), and the Association of Professors in Human and Medical Genetics. He serves as the AAPA external representative for genetics and genomics and is also a member of the Workforce Development and Optimization Committee for the American College of Medical Genetics and Genomics.

Patterson and his PA colleague, Laura Buch, MSPA, PA-C, of GGC's Greenville office have also coauthored two articles for the AAPA membership, 'A Day in the Life of a PA in Genetics' and 'Top 10 Things PAs Should Know About Genetics'.

In addition to his educational outreach for students and practicing PAs, Patterson also founded the Society for PAs in Genetics and Genomics (SPAGG), an organization dedicated to PA education in genetics and encouraging PAs to join this exciting and rewarding specialty.

Patterson worked at GGC as a laboratory technologist after completing his undergraduate degree and while deciding on his next career move. After completing PA school, he worked in family medicine before rejoining GGC as its first PA in 2018.

"As our understanding of genetics grows, we are also seeing the demand for genetics services growing rapidly," said Patterson. "The current workforce of MD clinical geneticists and genetic counselors is not sufficient to keep up with this demand. This is a wonderful opportunity to expand the workforce by including other advanced practice providers, such as PAs and nurse practitioners, to help with the workload."

While Patterson was GGC's first PA, the addition of PAs and nurse practitioners as part of the clinical team has grown in recent years with Buch practicing in the Greenville office as well as a new PA joining the Charleston office, and nurse practitioners in the Greenwood and Greenville offices.

"We are grateful for Wesley's leadership at GGC, both in the Division of Education as well as in the clinic," added Tribble. "His combined passion for patient care and education is a great benefit to GGC, to our patients, and to our future workforce."

BEST NAMED INTERIM CEO OF ACMG

Robert 'Bob' Best, PhD, a longtime GGC friend, collaborator, and recent fellowship graduate, has been named Interim CEO for the American College of Medical Genetics and Genomics (ACMG) and the ACMG Foundation.

Best's connection with GGC began when he joined the University of South Carolina School of Medicine (USCSOM) in 1986 where he was a Professor in Clinical Cytogenetics and Medical Genetics leading their clinical laboratories and supporting the genetic counseling training program. He currently serves as Professor of Biomedical Sciences with tenure at USCSOM-Greenville.

In 2022, Best completed an alternate track fellowship in Laboratory Genetics and Genomics through GGC's Medical Genetics Training Program and Harvard Medical School. He currently reviews GGC exome and genome data as part of his scholarly work at USCSOM- Greenville.

"I am excited about this opportunity and am immensely grateful for my association with GGC," said Best. "Much of what I think about the field of genetics has been influenced by my collaborations with the Greenwood Genetic Center."

GRANT FUNDS VIDEO PRODUCTION STUDIO

The GGC Foundation has received funding from the Fullerton Foundation to develop a videography program to enhance the Center's promotional and educational efforts.

The $100,000 grant will allow for the conversion of space in GGC's Genetic Education Center into a video production studio, as well as the purchase of cameras, lighting and other technical equipment, and the addition of a part-time videographer.

"This project will allow GGC to develop professional videos for a wide range of educational projects to enhance patient education, student career engagement, and continuing education topics in genetics for healthcare professionals," said Cady Nell Keener, Executive Director of the GGC Foundation. "We are grateful for the opportunity to enhance our current video library and promote the work of the Center with high-quality videos."

The new videographer will work closely with GGC's Foundation, Division of Education, and Office of Communications.

DUPONT HEADS REGIONAL GENETICS GROUP

Barb DuPont, PhD, Senior Director of GGC's Cytogenetics Laboratory, has been elected President of the Southeastern Regional Genetics Group (SERGG).

SERGG's mission includes enhancing and promoting the quality of genetic services in the Southeastern US. The organization provides a forum for exchange of information among professionals who provide genetic services and the consumers of these services.

"I am privileged and honored to be elected president of Southeastern Regional Genetics Group," said DuPont, who will serve a two-year term as President. "Working with other genetics professionals in the our region to promote clinical genetics and training in clinical genetics is an exciting opportunity."

Also serving on the SERGG Board of Directors from GGC are Julie Jones, PhD, GGC's Clinical Genomic Sequencing Program Director, and Laura Pollard, PhD, Lead Director of GGC's Biochemical Genetics Laboratory and Director of the Center's Laboratory Training Program.

Rare Rose II displays a genetic phenomena called phyllody where parts of the flower are replaced with leaf-like structures

RARE AND BEAUTIFUL

Artist and GGC mom shares message of compassion and acceptance through art

The lobby of the JC Self Research Institute at GGC has been transformed into a garden - of sorts.

The Center is hosting an art exhibit through December 15 entitled 'Rare Roses' that consists of 12 paintings that depict real roses with genetic variations. The series was created by Nicole Shannon, an artist from Greenville. Nicole was inspired by her son, who has a rare genetic disorder, and other individuals with genetic differences.

Quinn, now 4, was born with a myriad of health issues and complications. An ultrasound midway through Nicole's pregnancy identified the first glimpse that there may be a problem. After Quinn was born, genetic testing was performed and GGC's diagnostic laboratories identified a translocation between chromosomes 6 and 14 that resulted in the loss of 263 genes.

"Until I had a child with a DNA difference, I didn't know what it was like for people with genetic differences," shared Nicole in a presentation that she made about her family and her artwork to GGC employees as part of the Center's Gene Week in October.

"I was aware of how pervasive racism, sexism, classism, and religious prejudice are, but I never realized how dismissive people could be of other humans when they have a DNA variation. I've experienced many instances where my son has been treated as less than due to his DNA," she said.

"Each painting shows that we are all uniquely beautiful and deserving of love."

-Nicole Shannon

Nicole recounted her family's experiences from the time of Quinn's birth where people, even medical professionals, have acted "shocked that I unconditionally love my son and wanted what is best for him. They didn't see him as my son, my little boy, but as a freak - a mistake of nature."

That's when she first encountered Dr. David Everman, a clinical geneticist in GGC's Greenville office, who met with Nicole and her husband, Brendan, to discuss the genetic testing results.

"Thankfully, we eventually met Dr. Everman, and in a moment, everything changed," said Nicole. Everman, who retired from GGC in 2021, "was able to bring clarity to what caused Quinn's challenges. He was calm and shared only what he knew - without making assumptions of what he didn't know."

That was a turning point for Quinn's family. From that point on Nicole and Brendan became fierce advocates for their son - challenging the assumptions being made about his abilities.

After 56 days in the NICU, Quinn went home, just in time for Christmas with his family, without a breathing tube and without a catheter, thanks to persistence and advocacy by his parents.

The family soon travelled to Boston Children's Hospital where Quinn was accepted into the Center for Complex Care. He was evaluated by 27 specialists over the course of two exhausting weeks. He passed a swallow study and was cleared to eat, and they also initiated physical and occupational therapy.

Since that time, "Quinn has grown and improved in so many ways," Nicole said. "When we think back on what Dr. Everman told us - this was the possibility, this was the hope."

Nicole's experiences, and her unconditional love of her son, inspired her to find a way, through art, to express that every human is valuable and beautiful, regardless of their DNA.

She chose the rose, a symbol of beauty, because "roses are revered in almost every culture, and as living things, I was sure they must have genetic variation."

Nicole reached out to botanists all over the world who shared images of

roses that have genetic differences. "I was amazed by the photos and knew immediately that I needed to paint them."

Nicole painted twelve flowers with two different types of genetic alterations - the double flower and phyllody.

Five of the paintings are double flowers where the central reproductive organs of the flower are replaced by additional petals. The flowers appear fuller, and for this reason, many of the roses that are sold in floral shops and grocery stores actually have this genetic alteration.

The other seven paintings reflect phyllody, a genetic change that causes the reproductive organs of the flower to be replaced by leaf-like structures (as in 'Rare Rose 11' pictured left).

"Each painting shows that we are all uniquely beautiful and deserving of love," Nicole added.

GGC welcomed Nicole for an artist reception on October 6. Prior to the public event, she shared her very personal and moving story with GGC employees. She is donating 80% of the proceeds from the sale of Rare Roses to the Greenwood Genetic Center Foundation.

Nicole and her son, Quinn

"I can say without a doubt that Quinn would not be where he is today had the Greenwood Genetic Center not armed us with the knowledge and support we needed to help Quinn thrive."

"Dr. Everman provided calm, clarity, kindness, and support when everyone around us seemed overly anxious, emotional, and fearful," she added. "He undoubtedly changed the course of Quinn's care and with it, his life."

Learn more about Nicole and her Rare Roses series at nicoleshannon.com. You can also follow Quinn's progress on Instagram @OurMightyQuinn.

'Gene-erosity' Abounds at GGC

GGC was founded in 1974 through a major philanthropic gift from Jim Self. In the ensuing 48 years, GGC and the patients we serve have continued to benefit from generous donors who have supported all areas of the Center's mission.

Cady Nell Keener, CFRE, who joined GGC in January of this year has been personally touched by the many patient stories she has heard during her first year at the helm of the GGC Foundation.

GGC Foundation staff (L-R): Boo Ramage, Director of Philanthropic Services; Cady Nell Keener, Executive Director; and Amy Botts, Coordinator

"The work that is happening here at GGC is truly remarkable," said Keener. "I have had the immense privilege to meet so many GGC families and hear even more stories of how the compassion shown during clinical visits, the answers provided by diagnostic testing, and the hope generated from promising research have changed lives." Keener has made a point of inviting some of those families to share their experiences with the GGC Foundation Board of Trustees during their quarterly meetings to help them understand the impact of the Center's work.

"During this holiday season, which is so much more about giving than receiving, we invite you to consider sharing your compassion and your dollars to support initiatives such as the GGC Cares Fund, which helps to offset the cost of genetic testing for patients who lack adequate insurance coverage," said Keener. "And while your gifts make a huge impact, I often find that the donor receives the greatest blessing. As author Randy Alcorn says, 'Giving infuses life with joy.'"

To support the GGC Cares Fund, use the enclosed envelope, visit www.ggc.org/foundation/donate or scan the QR code.

SCAN ME

Donations to the GGC Foundation can also be made through gifts of stock, IRA roll overs, or legacy estate planning. For more information, please reach out to Keener at the GGC Foundation ckeener@ggc.org or (864) 388-1813.

Race the Helix-Greenwood

GGC patient is overall race winner

JT Shorter loves to run.

And he's very good at it. That was evident with his first place finish at GGC's 12th annual Race the Helix-Greenwood, held during GGC's Gene Week festivities in October.

But this wasn't the first time that JT had been to GGC. His first visit was at age nine when he was referred for a genetics evaluation following his diagnosis of autism spectrum disorder.

"We suspected that James Thomas (or JT as he likes to be called at school), might be on the spectrum around age 3 or 4," said his father, KJ Shorter. "He was always very verbal and does well academically, so it can be difficult for some to understand that he has this diagnosis, but he has social struggles, anxiety, and sometimes might appear rude when he doesn't mean to be."

His parents shared that JT has learned to deal with some of these struggles through ABA, or Applied Behavioral Analysis therapy at Project HOPE Foundation in Greenwood, and he is thriving at Thornwell Charter School in Clinton where he runs on the cross country team.

"His school has a wonderful faculty and staff who understand that all children are different, and they meet them where they are," said KJ.

JT was only able to run one meet during his first year of cross country because of COVID, but he ran in 8th grade and again this year as a high school freshman.

"Running has been wonderful for him," said his mother, Kristen. "It has further developed his social skills, and he's becoming a leader on the team by encouraging his teammates. It also helps to manage his excess energy."

The Shorter family are regulars at Race the Helix events. They served as the host family for the 2018 Greenwood race, presenting the awards and sharing their family's GGC experience with JT, his 8-year-old brother, Knox, and six-year-old brother, Elias, all of whom are GGC patients.

"We are very thankful for the Greenwood Genetic Center and want to let people know about the wonderful resource we are blessed to have in our state," added KJ.

The Shorters are also close friends with the Shenal family who started Race the Helix in 2010. Ryleigh Shenal was one of Knox's first friends when he started therapy at 6 months, and 'he was always so excited to see Ryleigh every time he went to therapy," said Kristen.

"The passion that the Shenals share about the Greenwood Genetic Center with all who will listen is inspiring, and we genuinely wanted to be part of the race," added Kristen.

Race the Helix - Greenwood is in its 12th year of raising awareness and funds to support GGC. All proceeds from this year's event benefit the GGC Foundation's 'GGC Cares Fund' which helps offset the cost of genetic testing for patients who are uninsured or under insured.

"The 2022 event was our largest Race the Helix ever," shared Cady Nell Keener, CFRE, Executive Director of the GGC Foundation. The Greenwood race attracted over 320 participants, both in person and virtually, and raised over $50,000 in sponsorships.

One of the presenting sponsors, Bionano, a provider of genome analysis solutions based in San Diego, not only sponsored the race, but they also held their own Race the Helix event in California for employees to participate together. Bionano President and CEO, Erik Holmlin, PhD, and Chief Medical Officer, Alka Chaubey, PhD, even flew to Greenwood to participate in person with Holmlin winning his age group.

"We are so grateful to all of the participants, volunteers, and generous sponsors who made this event such a success," added Keener.

JT Shorter, 14, approaches the finish line as the overall winner of the 2022 Race the Helix-Greenwood

GGC Director, Steve Skinner, MD, with Holmlin and Keener

The Leary Clinic

Family honors memory of sister

Dianne Leary was a strong, intelligent and persistent woman - who happened to have a genetic disorder.

Following Dianne's death from Charcot-Marie-Tooth disease in 2020, her siblings, Julius Leary, MD, Kathie Marsh, and Lynne Lovett, created the Dianne Patricia Leary Fund for Neuromuscular Disorders at GGC to support research into this rare group of genetic conditions.

In October 2022, during GGC's annual Gene Week celebration, the Center formally recognized this gift and dedicated 'The Leary Clinic' in Greenwood in memory of Dianne.

"It was an honor and privilege to have shared life with Dianne. She was an inspiration to all who knew her, " said Dr. Leary at the dedication ceremony. "The Leary Clinic was designed as a symbol of hope for patients who have any genetic neuromuscular disorder - hope for a diagnosis, hope for treatments, and hope to make their lives more comfortable."

In addition to Dianne's family and friends, Aaron Baker and his son, Brody, were also in attendance at the clinic's dedication. The Bakers traveled from Atlanta, Missouri to be part of the celebration. Brody, 12, also has CMT, though he has a different type than Dianne.

When Brody's genetic testing revealed a variant of uncertain significance in the *AIFM1* gene, which causes an ultra-rare form of CMT, the Bakers were looking for answers.

That answer came when Aaron Baker reached out to GGC in 2020 after seeing a press release about the Leary Fund. GGC's Director of Research, Rich Steet, PhD, who had previously worked on the *AIFM1* gene, was able to study some of Brody's skin cells and confirm that his variant was disease-causing.

And through the Leary Fund, the Bakers and GGC researchers are developing a possible project to not only understand why Brody's symptoms are milder than others with his form of CMT, but also to identify possible treatment targets to improve the quality of life for Brody and for others with this neuromuscular condition.

"We are indebted to the research team at GGC for helping us better understand Brody's genetic change, and we are excited about the possibility of working with the Center on future projects to identify potential therapies," said Aaron Baker. "Our family is grateful for the Leary's gift that has made much of this work possible."

Brody himself also spoke at the dedication ceremony, sharing, "Not all diseases are easy to see. This research will help myself and others like me hopefully have hope for a cure down the road."

Photo: L-R: Rich Steet, PhD, Brody Baker, Aaron Baker, Kathie Marsh, Julius Leary, MD, Lynne Lovett, and Steve Skinner, MD with a photo of Dianne Leary at the dedication ceremony

WELCOME GGC' S NEW FACULTY

Emily Black, MD, joined GGC as a clinical geneticist in the Center's Greenville office. She is board certified in pediatrics, medical genetics, and medical biochemical genetics. Dr. Black sees patients in Greenville's general genetics and metabolic clinics

Will Burns, MD, has returned to GGC as a clinical geneticist in the Columbia office. He completed his medical genetics residency at GGC in 2021. Dr. Burns sees general genetics patients in Columbia and metabolic patients in both Columbia and Greenwood

Aneta Kaczmarczyk, PhD, joined GGC as Assistant Director of the Biochemical Laboratory. Dr. Kaczmarczyk oversees biochemical testing, signs out patient reports, and implements new testing technologies.

Natasha Rudy, MS, CGC, joined GGC's Greenville office as a clinical genetic counselor. She provides general genetic counseling for pediatric and adult patients in GGC's Greenville office.

Brett Sparks, FNP-C, joined GGC's Greenwood clinic as a nurse practitioner. She provides care for patients through GGC's general genetics clinics.

Tonya Moss, research technologist and coauthor on the paper, loads a Western blot to analyze the P5CS enzyme encoded by the ALDH18A1 gene.

THE HOW

Why understanding *how* a gene functions is critical to the development of effective treatments

The Division of Research at GGC is tasked with asking, and then answering, some of the most challenging questions in biology.

"The questions are often posed to us by our colleagues in the clinical and diagnostic lab divisions when they identify a patient with atypical symptoms or find a novel or unusual genetic variant," said Rich Steet, PhD, Director of Research at GGC, pictured below. "It then becomes our job to try and figure out how the gene variants in question cause the symptoms that they do. Once we get to the 'how this happened', we have a better sense of the possible ways the condition may be treated."

"Once we get to the 'how this happened', we have a better sense of the possible ways the condition may be treated."

-Rich Steet, PhD, Director of Research

Steet and his research team recently collaborated with colleagues at the University of Georgia (UGA), Johns Hopkins School of Medicine, and Children's Hospital Colorado, on another 'how' for two GGC families who have a genetic variant in the gene *ALDH18A1*.

Photo: Clemson University

Variants in this gene are known to cause a spectrum of disorders that include skin and neurological findings. *ALDH18A1* has also been implicated in several human cancers including breast cancer and melanoma.

In one GGC family, a ten-year-old girl with significant developmental delay, neurologic issues and gastrointestinal concerns, was found to have the new variant.

In another family, three siblings who all have the gene variant, exhibit developmental delays, growth issues, loose skin, and gastrointestinal symptoms.

"At this point, all that we can do to treat these children has been to manage some of their symptoms," said Mike Lyons, MD, Director of Clinical Services at GGC. "If we can understand how the underlying disease is caused by the genetic changes, we have a better chance of intervening at the most basic level and improving their overall health and development."

Steet and colleagues used skin cells from one of these four patients to, first of all, confirm that these new variants that were identified were truly disease-causing.

"Using cell-based experiments, we were able to show that these genetic variants impact the function of the enzyme encoded by the *ALDH18A1* gene," said Steet, "but that work did not provide any insight about how the genetic change was causing the specific symptoms seen in the patients."

For that, the team explored how the enzyme that is encoded by this gene, P5CS, was altering the metabolic and transcriptional program of the patient's cells.

"Our colleagues at UGA looked at the metabolomic profile which is basically a large-scale study of all of the small molecules or metabolites with the cell," said Steet. "What we found is that the defective enzyme was affecting numerous metabolic

pathways that had not been identified in prior studies."

When the P5CS enzyme is impaired, as it is in these two families, there is a reduction in several metabolites, including a key component of collagen, which Steet said may explain the loose skin findings, and an antioxidant molecule called glutathione which helps protect against oxidative damage and may explain the neurological symptoms.

The researchers also looked at gene expression effects using RNA sequencing, uncovering other sensitive pathways that may contribute to the skin and neurological findings in these children.

"The data from this project point to the involvement of antioxidant responses in the disease process," said Steet. "We believe that antioxidant supplementation, or drugs that boost the production of antioxidant molecules, may be a possible therapeutic avenue."

Sneha Mokashi, PhD, a postdoctoral associate in the Steet lab and coauthor on the paper, reviews gene expression data for ALDH18A1.

Further studies on cells and animal models to test this hypothesis will be needed before any treatment plan for patients can be considered.

This work was published online in September in advance of print publication in the journal, *Human Molecular Genetics*.

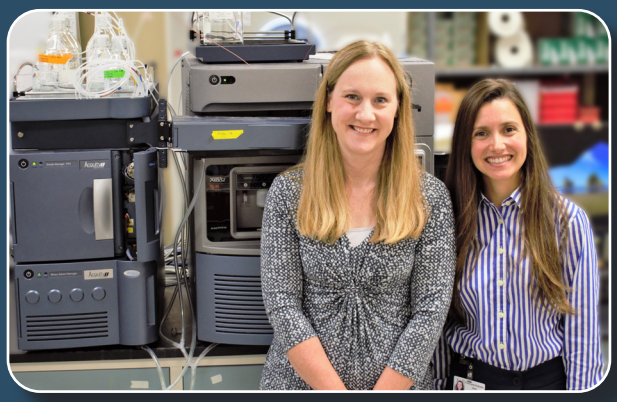
Biochemical Lab Earns Grant Funding

MPS Society Funds GGC Study on the Neurological Impact of Treatment

Mucopolysaccharide (MPS) disorders are a rare group of conditions caused by an enzyme deficiency that prevents the cells from breaking down large sugars into smaller components that can be recycled. The accumulation of these sugars, called glycosaminoglycans (GAGs), leads to a damaging buildup in the cells. A patient's symptoms will vary depending upon exactly which GAG is in excess, but often include both physical and neurological symptoms.

MPS disorders are typically diagnosed by analyzing the amount of GAGs present in the patient's urine or blood. Current treatments for MPS disorders involve the use of a synthetic enzyme to replace the one that the patient is missing or a stem cell transplantation. Several clinical trials are underway for novel therapeutic approaches for MPS.

"When the treatment is effective, we see a decrease in urine and/or blood GAGs, showing us that the treatment is working," said Francyne Kubaski, PhD, staff scientist in GGC's Biochemical Genetics Laboratory. "That's called a biomarker - it's a quantitative measurement of how well a treatment is working, or if it's working at all."



Biochemical Laboratory Director, Laura Pollard, PhD, and Staff Scientist, Francyne Kubaski, PhD, with the mass spectrometry instrument that measures GAGs in the CSF.

Kubaski adds, "However, because MPS disorders often have neurological symptoms, and it's harder to get that drug into the brain, measuring GAGs in the blood or urine may not accurately reflect the impact of the treatment on the brain."

To determine if treatment is reaching the brain, GGC's Biochemical Lab was awarded a $50,000 grant by the MPS Society in June for a yearlong project.

"We are using two different methodologies to measure GAG levels in the cerebrospinal fluid (CSF) of patients who have an MPS disorder as well as pediatric and adult controls. CSF is the fluid that bathes the brain and spinal cord, so it more accurately reflects the impact of the disease and treatment on brain cells," said Laura Pollard, PhD, Director of GGC's Biochemical Genetics Lab and lead scientist on the project. "By using two different methods, we can compare the results and understand the strengths and limitations of each method. This data will then be made available to the MPS research community to advance pre-clinical studies and clinical trials."

LAVERDURE SELECTED TO MOBILE LAB BOARD

Ashley LaVerdure, Operations Manager and Instructor for the Greenwood Genetic Center's Division of Education, has been selected to serve on the Board of Directors of the Mobile Laboratory Coalition.

The Mobile Laboratory Coalition (MLC) is an international community of traveling STEM outreach programs. The organization provides education, fosters collaboration, and serves as a central resource of knowledge, curricula, and best practices for member programs.

LaVerdure, who will serve a two-year term on the MLC board, joined GGC in 2019 as an instructor traveling across SC and leading activities for middle and high school students through the Center's popular 'Gene Machine' and 'Helix Express' Mobile Science Labs. She was recently promoted to operations manager for the program.

"I'm thrilled to serve on the MLC board to continue to encourage collaboration within our community and to learn from other programs dedicated to informal science education," said LaVerdure, who attended the 2022 MLC meeting in Washington, DC in July. "Mobile lab programs face unique challenges, and working closely with others in the mobile lab community benefits everyone – most importantly, our students and teachers."

"We are so proud that Ashley will be representing GGC as a leader in this coalition that has been so beneficial to the development and success of our mobile lab outreach here in South Carolina," said Leta Tribble, PhD, GGC's Director of Education. "Ashley is a wonderful instructor with initiative and great ideas on how to expand our reach. GGC and the MLC are fortunate to benefit from her enthusiasm and leadership."

Ashley, who has a degree in Biology and Education from NYU, is passionate about science education, "Throughout my time at GGC, I have developed a deep love for informal STEM education, and I'm so grateful to be part of this field. Being able to witness my students' a-ha moments has brought me intense fulfillment."

EDUCATION EXPANDS OFFERINGS

GGC's Junior Genetics Scholars Camp in Greenwood has always been a popular summer activity for area high school students. Since 2018, GGC's Division of Education has attracted science-minded students to GGC's Greenwood campus for hands-on lab experiments, direct access to professionals in the field of genetics, and lots of fun activities.

Kirah Jones works on a microscope activity at the Myrtle Beach camp.

In 2022, GGC's education team partnered with Horry-Georgetown Technical College (HGTC) to offer a second session of the week long camp in Myrtle Beach. The Greenwood and Myrtle Beach camps served a total of 19 students who studied topics such as CRISPR gene editing, the genetics of behavior, and bioinformatics.

"For five years, the camp has been a highlight of our summer, and we are excited to be able to provide these engaging experiences to students in other parts of our state who aren't able to make the trip to Greenwood," said Leta Tribble, PhD, Director of Education. "We are grateful for the partnership of HGTC, and we hope to expand the camps even further in future years to provide these fun opportunities in other locations."

Also, during the school year, the outreach team conducts overnight trips each month to serve schools that are too far from Greenwood for a day trip. This year, they have started offering STEM-based activities at afterschool programs, such as Boys and Girls Clubs, in these areas to share their passion for genetics with younger students or with those who may not have the opportunity through their classes at school. "We have received a very positive response from these afterschool programs who are excited to have a unique and engaging activity for the children they serve," said Dr. Tribble. "We are excited to see our programs growing."

GGC - MUSC Working Groups Underway

With the GGC-MUSC membership agreement finalized earlier this year, several working groups have begun addressing joint initiatives between the two organizations. Under the initial plan of the membership agreement, five working groups have been established with joint leadership and membership from both GGC and MUSC.

"The initial stages of these working groups have been productive, and we are working to build relationships and strengthen connections across the GGC and MUSC," said Steve Skinner, MD, Director of GGC. "We are excited about the progress and plans of each group to improve access to genetics care for patients and families across SC and to strengthen both of our organizations."

Workgroup A, led from GGC by Mike Lyons, MD, Director of Clinical Services, is tasked with increasing access to genetics services. This group's initial goals are focused on improving access to care by coordinating referrals and appointments, more efficient use of genetic counselors, and expanding the use of telemedicine, including eConsults and eVisits. This workgroup is also coordinating workforce recruitment and utilization across both institutions.

Workgroup B, led from GGC by Mike Friez, PhD, Director of GGC's Diagnostic Laboratories, is working toward aligning and optimizing laboratory testing with the goal of retaining genetic testing in SC by increasing laboratory referrals to GGC and improving patient and provider access to genetic testing at both institutions. This workgroup is working to reduce the costs of genetic testing and improve the ease of ordering testing for providers.

Dr. Friez also leads Workgroup C which is tasked with investing in innovative approaches to clinical delivery, genetic testing, and research. This team is driving collaboration to strengthen the use of shared technologies and key expertise, as well as developing new tests and products to support patient care.

Workgroup D, headed by Paul Pridmore, GGC's Chief Operating Officer (COO), seeks to optimize processes such as electronic medical records, reporting capabilities, billing, and other supportive processes to increase efficiencies and decrease costs. Kevin Farren, GGC's Director of Data Integration and Management, is leading the effort from GGC to integrate electronic medical records and laboratory information management, a critical first step in increasing efficiencies in genetic services.

Workgroup E consists of the Joint Operating Committee which includes Skinner, Pridmore, and Brandi Buff, GGC's Chief Financial Officer (CFO), along with MUSC Health's CEO, Pat Cawley, MD; CFO, Lisa Goodlett; and COO, Tom Crawford, PhD. This committee is tasked with providing resources and operational support to enable the success of the workgroups and to ensure the sustainability of the initiatives from the workgroups.

"Throughout the early months of our work together, we have seen wonderful collaborations between GGC and MUSC faculty and among our newly-appointed board members," added Skinner. "The possibilities for the growth and improvement of genetics services are significant, and patients and families across SC will reap the greatest benefits."

First meeting of the new GGC Board of Directors:
L-R Seated: Dell Baker and Holisa Wharton, PhD; Standing Chris Przirembel, PhD, Steve Skinner, MD, Jay Nexsen, Charles Schulze, John Miller, Bill Stevens, Howell Clyborne, Terri Barnes, and Pat Cawley, MD.
Not pictured (joined virtually) Fritz Butehorn, MD, Richard Christian, MD, and Reid Conrad.

EMPLOYEE UPDATES

Congratulations to the following GGC employees who have excelled in their roles and have been granted promotions in recent months...

Jamie Butler was promoted to a supervisory role in the Diagnostic Laboratory

Raymond Caylor, PhD, was promoted to Associate Director of the Molecular Diagnostic Laboratory

Jessica Cooley Coleman, PhD, was promoted to Staff Scientist in the Molecular Diagnostic Laboratory

Katy Drazba, MS, CGC, has been named Interim Lead Genetic Counselor

Brittany Hennigan was promoted to Lab Manager in the DNA Diagnostic Laboratory

Ashley LaVerdure was promoted to Operations Manager for the Division of Education

Olivia Nail was promoted to Curriculum Specialist for the Division of Education

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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