Greenwood Genetic Center

Winter 2021

A Newsletter for the Friends of the Center

Everything you need to know about eVisits

Using technology to improve access to genetics care

p. 2

Where

COMPASSION

p. 4

Persistance and technology combine to provide long-awaited diagnoses

INSPIRES

p. 7

The Gene Machine and Helix Express are back on the road and in classrooms

PROGRESS

p. 8

Research and diagnostic labs collaborate to clarify uncertain newborn screening results

eVisits - A New Option for Patient Care

Your questions answered...

The Greenwood Genetic Center, with funding support from The Duke Endowment, is developing new protocols to improve access to genetic services for patients across South Carolina. Part of this initiative includes the addition of a new type of patient visits called eVisits.

"We are excited to be expanding our clinical visit offerings to better meet the needs of our patients," said GGC's Director of Clinical Services, Mike Lyons, MD, who is leading the project. "eVisits are a new concept for many patients, so we wanted to provide some answers to commonly asked questions to help patients feel more comfortable with this new care option."

Q) What is an eVisit?

A) An eVisit, or electronic visit, is a way to use technology to provide genetics care for patients at their convenience.

Instead of scheduling an appointment to come into the office or a set date and time to connect for a telemedicine appointment, an eVisit allows patients and providers to connect on their own time to share information, updates, and recommendations.

Q) What's the difference between an eVisit and telemedicine visit?

A) eVisits and telemedicine visits both use technology to improve access to genetics providers.

In a telemedicine visit, you will schedule a date and time to meet with the provider through a video link. The patient will need to be available during that specific time, and the family will be able to speak directly with the provider via video. A limited physical examination may also be performed through telemedicine.

An eVisit does not require a specific appointment. You will communicate with your provider through a secure online site by sending information, answering their questions, and asking your own questions. The provider will review the information you provide and respond within two business days.

Q) How does an eVisit work?

A) If your provider determines that an eVisit is appropriate for you or your child, that option will be offered to you when it's time to schedule your follow-up appointment.

If you choose to have an eVisit, you will communicate with your GGC provider through a secure, HIPAA-compliant online portal. Your provider will send questions for you to answer at your convenience. They may also request that you upload other information such as recent photos. They will review your answers, and within two business days, they will respond with their comments and recommendations. You can respond with additional questions. The entire exchange will take no more than seven business days.

Q) What are the benefits of an eVisit?

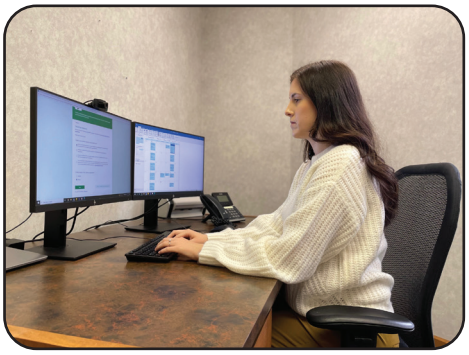
A) eVisits allow for patients to check in with their provider, share information, and ask questions without having to schedule a specific appointment. It can be done at your convenience and does not require travel, taking time off of work or school, or finding childcare for other children. eVisits can also usually happen more quickly. You do not have to wait to get an appointment like for telemedicine or in-person appointments.

Q) What are the challenges of an eVisit?

A) eVisits may not be ideal for new patients or for any patient who requires a physical exam. They may also not be appropriate for patients who have many complex questions or for diclosure of abnormal genetic testing results. eVisits do require an internet connection, so they will not work for patients who do not have this access.

Q) What if I or my child needs additional genetic testing? Would an eVisit work?

A) Yes, in many cases. After communicating with you through an eVisit, if your provider suggests additional genetic testing, they will discuss that with you through the portal. For many tests, genetic testing can be performed using a saliva sample. GGC can have a collection kit mailed to your home where you can collect the sample and mail it back to GGC using postage-paid packaging.
In some cases, testing may require a blood draw. In those situations, you would need to come in to a GGC office or another lab to have the sample drawn.



Cover: Mike Lyons, MD, Director of Clinical Services (seated), Brian Albon, Clinic Operations Manager, and Mattie Sullivan, Genetic Assistant, are part of the GGC team implementing eVisits.

Above: Sullivan reviews a patient's eVisit requests and assists with scheduling.

Q) Are eVisits right for all patients?

A) No. In certain circumstances, it is preferred to have an in-person or telemedicine visit, especially for new patients. At this time, GGC is only offering eVisits for patients who have been previously seen and are scheduled to have a follow-up visit with one of our providers. eVisits do not allow for a full physical examination. If your child has a confirmed diagnosis or is an established patient at GGC, your provider will determine if an eVisit may work for your follow-up care.

Q) How do I know if an eVisit is right for me or my child?

A) If you or your child are due for a follow-up appointment with GGC, your provider will suggest whether an eVisit may work for you. When reaching out to you to schedule your follow-up appointment, you will have the ability to choose between an eVisit, a telemedicine appointment, or an in-person visit.

If you are an existing patient and have more questions about eVisits, please contact the GGC office where you were seen.

LYONS NAMED SC TELEHEALTH PIONEER

GGC's Director of Clinical Services, Mike Lyons, MD, was honored as the 2021 SC Telehealth Pioneer by Palmetto Care Connections at the 9th Annual Telehealth Summit in November.

The award recognizes an individual champion in the areas of clinical care, education, or policy who implemented telehealth within a practice, health center, school-based or administrative setting.

Lyons was nominated for the award for his work in instituting telehealth at the Greenwood Genetic Center in 2016. The program allowed GGC to more easily transition to fully virtual care in the early days of the pandemic.

He has also been active in research, demonstrating high patient satisfaction and comparable diagnostic yields between virtual and in-person visits. Lyons is a vocal proponent for telehealth, and has partnered with numerous organizations including MUSC and the SC Telehealth Alliance to advance telehealth opportunities in SC. He also leads a GGC project funded by The Duke Endowment to institute eVisits (p.2) and eConsults, utilizing technology to improve access to genetics care for all across SC.

KAY RECOGNIZED AS SC'S HR RISING STAR

Talitha Kay, Assistant Human Resources Manager at the Greenwood Genetic Center (GGC), has been named the 2021 HR Rising Star by the SC Chamber of Commerce. The statewide award recognizes an individual with fewer than 10 years of professional experience in human resources who has already made an impact.

Kay is a 2014 graduate of Lander University with a degree in Business Administration with an emphasis in Healthcare Management. She worked at GGC as a college intern and joined the Center as a Human Resources Generalist upon graduation. She was promoted to Assistant Human Resources Manager in 2020. She has also served as President of the Piedmont Area Human Resource Association (PAHRA).

“Talitha truly embodies the epitome of a human resources professional – working within our organization and the larger profession to improve our workplace and support our workforce every day," said Steve Skinner, MD, Director of GGC. "We are fortunate to have her on our team!”

The award was presented at the statewide meeting of the Society for Human Resource Management (SHRM) in Myrtle Beach in September.

FLANAGAN-STEET IS WOMAN OF EXCELLENCE

GGC's Director of Functional Studies, Heather Flanagan-Steet, PhD, was named by the Greenwood Chamber of Commerce's Women's Leadership Council as their 2021 Woman of Excellence. This award honors a local woman who has started a community or business initiative, contributing time, talent, and resources, and utilizing her influence to enhance opportunities for women. The award was presented at a Chamber event in December.

Flanagan-Steet relocated her zebrafish laboratory from the University of Georgia to GGC in 2018, in large part through a desire to have a more direct impact on patient care. "Her research has led to many families finally receiving an accurate diagnosis and several breakthroughs that are moving patients closer to a treatment for their rare disorder," said Steve Skinner, MD, Director of GGC.

Flanagan-Steet has also been involved in SCBio, a statewide organization that promotes the life science industry. She has spoken at their annual conference and participated in a ‘Women in Science’ roundtable discussion with other female leaders hosted by Lt. Governor Pamela Evette. She is also an active member of the Center's Inclusion and Diversity Advisory Council.

Cadence Post (left) and her mom, Connie Post

NEVER GIVE UP!

Novel Technologies Combined with Persistance Leads to Long-Awaited Answers.

Frustrating - that's one way families often describe the diagnostic odyssey that is all too frequent for patients with a suspected genetic disorder.

Not having an answer is difficult - not understanding what caused your child's health and developmental challenges, not knowing if it could recur in the family, or if there's something you could be doing differently that would help.

But with the current climate of rapidly expanding genetic technologies, answers are finally being discovered for many families.

Cadence Post, 19, has been a patient at GGC since age seven. She was diagnosed with autism spectrum disorder after a period of developmental regression which included loss of speech.

"At the time we didn't know very much about autism and we certainly didn't know why she had it," said Cadence's mother, Connie. "She was later diagnosed with an intellectual disability, but she also had other things going on that there were no answers for."

In the 12 years she has been part of GGC, Cadence has undergone numerous genetics tests and has been involved in GGC's autism research studies, none of which provided an answer of why - that is until recently.

"A few years ago they asked if they could bank her DNA, so they could continue to study her case," said Connie. "And then last summer out of the blue, Dr. Roger Stevenson called to tell me that there was a new technology that allowed them to search her entire genome at one time. They found that she had a very rare genetic disorder that only 250 people in the world have. And as he explained what it was, it answered all the questions that we have had over the past 15 years."

"For years, the genetic center was searching for answers - working faithfully behind the scenes, fighting for families who needed answers to the hard questions."

- Connie Post, mom of Cadence

Dr. Stevenson ordered two new tests on Cadence's banked DNA. This first one to come back was EpiSign, which showed that Cadence had a pattern of gene expression that was consistent with a rare disorder called Tatton-Brown-Rahman syndrome. Cadence's whole exome sequencing was pending when EpiSign came back, but a couple of weeks later, that testing confirmed a mutation in the gene *DNMT3A*, which is known to cause this rare disorder.

"I think that every parent who has a child who suffers asks themselves if there's anything they could have done differently for a different outcome, even if they never say that out loud," admitted Connie. "As Dr. Stevenson explained the test results, he let us know that this is the reason she had autism, and ID, why she was so tall, the way that she looks, and so many other things that did not fit for the past 15 years. Then he spoke straight into my heart as a mother, and said 'Mrs. Post, there's nothing that you could have done differently to keep this from happening,' and he gave me relief that I didn't even know that I needed."

Connie added, "For years, the genetic center was searching for answers - working faithfully behind the scenes, fighting for families who needed answers to the hard questions."

Parker Egbert's family began their diagnostic odyssey with GGC when he was just three years old.

"At the time, autism wasn't that prevalent, and I was grasping for explanations of why Parker was Parker. Blood, urine, dad getting tested, me getting tested, sisters getting tested and still no answers," said Parker's mother, Laura Egbert.

"I always told people that a test or diagnosis didn't change Parker or the amount we loved him but I secretly hoped and prayed for answers. A

diagnosis was honestly all we ever prayed for."

Now 18 and a member of the US Paralympic swimming team, Parker's speech, occupational, and ABA therapy have proven very beneficial, and he is soon to graduate from high school. But only recently did the Egberts receive an answer for Parker's challenges.

"Every single year for almost 15 years, we came, met with doctors and ran more tests," recalled Laura. "Still, no answers… then in January of 2021, we hit the jackpot and finally received a diagnosis for our sweet boy."

GGC physician assistant, Wesley Patterson, had ordered a whole exome sequencing analysis using Parker's whole genome sequencing data (see below). The results revealed a *de novo* alteration (meaning that it was new to Parker and not present in either of his parents) in a gene called *POLR2A*. While Parker's specific genetic change had never been reported before, similar changes in this gene are known to cause an autosomal dominant neurodevelopmental disorder with intellectual disability that fit with Parker's clinical issues.

Parker Egbert in Tokyo for the Paralympic games in 2021. Inset: Parker with his sisters Lucy (left) and Chloe (right)

"When you come to the Greenwood Genetic Center, they become your family. It is like a team of people (parents included) all looking for answers and ways to benefit their child," added Laura. "GGC has been that for us - family. I'm not sure who was more excited about his diagnosis, Wesley or me!"

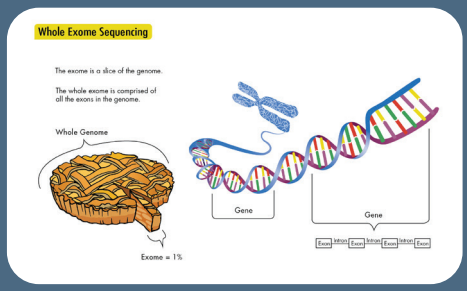
While neither answer is able to provide a targeted treatment or cure for Cadence or Parker at this time, the answers have been welcomed by their families.

"I was nervous to come to the genetic center. I was scared of what I might find out. I was scared we wouldn't find anything either," said Laura. "In the end, Parker's pediatrician told me knowledge is power. Those words still ring true, knowledge IS power. We have learned so very much from Parker, how to love unconditionally, how to encourage, how to have patience and just how amazing kids like Parker are!"

Exomes, Genomes, and Methylation Explained

New laboratory technologies finding challenging diagnoses

A genome is the complete set of genetic material present in an individual. The human genome consists of approximately 20,000 genes that encode proteins, along with DNA that has other functions such as regulating the expression of those genes (turning them on or off, up or down). Whole exome sequencing (WES), which became clinically available at GGC in 2014, allows for the analysis of the protein coding region of the DNA, also called exons. The exome is the part we understand the best, but it only comprises around 1-2% of the entire genome. WES has improved the ability to diagnose patients by allowing for analysis of changes in genes across the genome rather than a gene-by-gene approach.



Then, in 2019, GGC partnered with London Health Sciences Centre in Canada to begin offering EpiSign, a novel test that looks for patterns in methylation, a chemical tag that helps control gene expression. There may not be a mutation within the gene itself, but a change somewhere else in the DNA that causes the normal gene not to be expressed appropriately. There are currently more than 50 genetic disorders that can be identified through EpiSign, many of which are very difficult to diagnose using other methods.

Now, GGC has the capability to sequence a patient's entire genome. Using the NovaSeq 6000 sequencer, the diagnostic lab can generate billions of data points on a patient that are analyzed through complex algorithms to identify genetic changes that match that patient's specific clinical features. The current clinical testing, called WES-XL, analyzes the whole exome plus other genetic changes that can provide answers that prior testing may have missed. By running patient samples on what is termed a 'whole genome backbone,' the patient's complete genomic data is available to be analyzed.

"Once the sequencing has been done, we can go back to that data to analyze it over and over as needed. The patient's data doesn't change, but our understanding of genetic variants changes frequently as we learn which variants are benign, which are disease-causing, and how those variants contribute to disease," said Julie Jones, PhD, GGC's Clinical Genomic Sequencing Program Director. "This technology allows us to run a patient sample once and continue to assess what the data means for years into the future as our understanding grows."

GGC, MUSC expanding relationship

Organizations Sign Letter of Intent

GGC and The Medical University of South Carolina (MUSC Health) have signed a letter of intent to expand their longtime collaboration to improve access to high-quality, coordinated and cost-effective genetic services and their recent affiliation to drive innovation in the genetics field. A definitive agreement and approval by both organizations' boards is still required and anticipated by the end of the year.

The deeper relationship between GGC and MUSC Health presents opportunities to further improve access and enhance patient care, increase access to a broader range of educational opportunities for MUSC students, and position the organizations to take advantage of future strategic research initiatives. The two entities have worked together on clinical consultations, provider education, and research for more than a decade. According to MUSC, adding to the depth and breadth of the almost two-year old formal affiliation with the state's most advanced and innovative genetic center was an easy choice.

"We are so pleased to continue aligning and innovating with this like-minded and advanced care provider for the benefit of the state's citizens," said James Lemon, D.M.D., chairman of the MUSC Board of Trustees. "We are excited for what the future holds as we move forward together."

MUSC Board of Trustees vice-chairman and Greenwood resident, Charles Schulze, agreed. "I live in Greenwood, and I've said for years that a lot people don't understand what an absolute gem GGC is. They've helped about 100,000 families across the state make incredibly important decisions, discovered difficult-to-diagnose conditions, and have been there for these families every step of the way."

With recent and rapid growth in the understanding of how genetics impacts health throughout the lifespan, access to genetic information is increasingly important for individuals to make informed healthcare and lifestyle decisions. With a primary goal of improving access for patients and their families, this expanded relationship between GGC and MUSC aims to leverage both organizations' strengths and expertise. Together they will provide high-quality care and access to the latest technological advances in diagnostics, research, and treatment. In the interest of better serving these needs, the expanded goals of the relationship include:

- Co-developing a strategic plan for genetic services.
- Continuing to increase access to clinical genetic services for MUSC patients and all South Carolinians.
- Building on collaborative telehealth platforms to improve wait times for appointments and consultations.
- Sharing critical resources and expertise where possible to lower costs.
- Pursuing additional workforce development, research, clinical trials and treatment collaborations.
- Advancing precision health and jointly serving as leaders in this innovative, dynamic area of health care.

Nearly every child in South Carolina who has been diagnosed with a genetic birth defect, developmental delay or other hereditary disorder has already benefited from GGC expertise, due to the center's depth of care for children with rare conditions and commitment to new technologies and diagnostics. GGC, a nonprofit institute centered on research, clinical genetic services, diagnostic laboratory testing and educational programs and resources, is focused on compassionate patient care and innovative scientific advancement. This deepened relationship with MUSC will mean GGC can expand their purview to include additional adult genetics services to help serve patients with cancer, cardiovascular disease, and other conditions.

"The Greenwood Genetic Center places great importance on collaborations that improve the quality of care and benefit the patients and families we serve," said Steve Skinner, MD, GGC Director. "Over the past two years, our affiliation with MUSC has expanded projects such as telemedicine that have had a significant and lasting impact on access to genetics care. With the further expansion of this relationship, GGC can have an even stronger impact on patient care through a connection to MUSC's broad subspecialties network and clinical trial experience, while GGC can enhance MUSC's ability to provide pediatric genetics care and state-of-the art clinical genetic testing. It's a win-win for both institutions, but most importantly, this collaboration is a win for the people of South Carolina who need genetic care, information, and resources."

Greenwood Genetic Center
Where Compassion Inspires Progress

MUSC Health
Medical University of South Carolina

David J. Cole, M.D., FACS, MUSC president, echoed the benefits of the affiliation offered by Skinner, adding, "Two years ago, we started to align the national caliber genetic expertise of GGC with our outstanding academic medical faculty and specialty care providers," he said. "It's been making a real difference for our patients. We're moving health care forward for all, bringing the best of both organizations so that we can create opportunities for more South Carolina citizens to understand, plan for and manage their health and wellness. By further connecting our work and accountability to each other, MUSC and GGC stand poised to deliver on precision health and even better patient care, research innovations and unique learning opportunities for our students."

"The Greenwood Genetic Center and MUSC individually provide exceptional care to patients across South Carolina, each with their own unique areas of expertise," said Dell Baker, chairman of the GGC Board of Directors. "By further combining our strengths and building upon the other's needs, this expanded relationship between our organizations has South Carolina poised as a leader in providing the best and most advanced genomic medicine for its citizens."

On the Road Again!

For 18 months, from the start of the pandemic until the current school year started, GGC's Gene Machine and Helix Express vehicles have sat virtually unused. When schools closed in March of 2020 and through the 20-21 school year when virtual learning, masks, and plexiglass shields were the norm, outside visitors and hands-on learning weren't safe.

As schools have returned to a more normal environment, the schedule for the Greenwood Genetic Center's outreach vehicles is finally filling up. The Center's calendar is nearly at capacity with 114 school visits planned this year.

The Gene Machine, a 40-foot bus, is designed to be a science lab on wheels. Students board the vehicle in their school's parking lot where they are led through innovative genetics activities by a team of genetics instructors. The Helix Express is a cargo van that allows GGC to transport lab equipment for use in the classrooms.

The goals of the program are three-fold – to improve genetic literacy among students, to provide biotechnology experiences to students who may not have access to that equipment, and to introduce a variety of in-demand careers in the life sciences.

In the 2018-19 school year, nearly 13,000 students experienced the Center's outreach programs. Last school year that number dropped by 90%. GGC instructors taught a few Zoom classes, but it wasn't the same.

"The very heart of our program is the face-to-face interaction and hands-on activities," said Dillon Gary, Lead Coordinator and Lead Instructor for the outreach program. "Engaging students with science requires laboratory exploration, and our program provides them that opportunity. They are diagnosing patients and solving forensic mysteries using the same equipment that GGC scientists use every day. It's fun and empowering."

GGC also has two new instructors, Olivia Nail and Marie Smith, bringing a new enthusiasm to the program. "I was a student on the Gene Machine in high school, and that experience was so memorable," said Nail. "I'm thrilled to be able to engage and excite students about genetics the way this program inspired me."

"We are so happy to be back doing what we love – interacting with students and getting them excited about genetics," said Leta Tribble, PhD, Director of GGC's Division of Education. "We recognize that the pandemic is far from over, but with vaccines and appropriate safety measures in place, we can now safely get back into the schools."

Above: Instructor, Olivia Nail works with students at Scotts Branch Middle/High School on one of the first Helix Express visits since the pandemic began.

Below: GGC's Education Team with the Gene Machine (L-R) Leta Tribble, PhD, Olivia Nail, Dillon Gary, and Marie Smith

'The Genetics of Immunity'

Using appropriate safety protocols and hybrid learning, GGC's Division of Education was able to offer the very popular summer teachers' course and Junior Genetics Scholars Summer Camp for high school students in 2021.

Both the summer teachers' course and high school camp focused on the very timely topic of 'The Genetics of Immunity.'

Twenty high school science teachers from across North and South Carolina participated in the online didactic portion of the course and visited the Greenwood campus in June to complete a two day hands-on workshop.

For the summer camp, held at GGC in July, GGC welcomed 24 high school students from Georgia, North Carolina, and South Carolina for a week of lessons, labs, and fun.

GGC instructors developed several activities allowing students and teachers to delve into the genetics of immunity including simulating PCR COVID testing, modeling protein structures, and exploring immunotherapy. Rebecca McPherson of the Clemson Center for Human Genetics led a presentation and activity on the use of fruit flies as a model organism for studying immune and other genetic disorders.

"I enjoyed genetics camp because I got to hang out with other kids, make new friends, and do cool experiments," said Joshua Albon, 15, a sophomore at Emerald High School in Greenwood. "My favorite experiment was racing the fruit flies!"

GGC has offered the teachers course since 1994, and the camp started in 2018. "These offerings are always popular with our teachers and students and provide a fun way to expand their knowledge and offer some unique activities and topics," said GGC's Director of Education, Leta Tribble. "We are so grateful that we've been able to continue these events safely through the pandemic."

Above: High school students, Joshua Albon and Riya Zutshi, work with vials of fruit flies during the 2021 Junior Genetics Scholars Summer Camp.

TEAMWORK CLEARS UP UNCERTAINTY

Research and Diagnostic Labs Join Forces to Clarify Newborn Results

Newborn screening, also known as the heel-prick test, that is administered to all babies before they leave the hospital is quite literally a lifesaver. Babies in South Carolina are screened for 53 rare genetic disorders so that therapies or even life-saving treatments can begin as soon as possible. GGC follows hundreds of patients who are on some form of treatment for one of these disorders.

Newborn screening is exactly that, a screening test, which means that in order to identify all of the affected babies, sometimes unaffected infants screen positive. These results must be sorted out quickly, so that babies who are true positives can begin treatment immediately and for those who are not, their families can be reassured and stop worrying.

But as more and more genetic disorders become amenable to treatment, the correct identification of true positives is becoming more complex. In 2020, GGC was the recipient of a grant from the National Mucopolysaccharidosis (MPS) Society for a project to help distinguish true mutations from those that are benign variants in the *IDUA* gene, mutations in which cause the disorder Mucopolysaccharidosis type I (MPS I).

A blood sample is taken from the foot of all babies born in South Carolina to screen for potentially treatable genetic disorders.

"Because symptoms of MPS1 are not present at birth, not knowing the significance of the gene variants puts patients and families in a state of limbo, uncertain as to whether they should start therapy." -Rich Steet, PhD

South Carolina and many other states have recently begun screening for MPS1, a lysosomal storage disease also known as Hunter syndrome or Scheie syndrome. MPS1 is highly variable, but generally causes significant skeletal, cardiac, vision, and hearing issues as well as developmental delay. Even in severe cases, symptoms are not generally present at birth.

Identifying MPS1 patients as newborns provides more options for treatment at an early stage when symptoms can be prevented or lessened through treatments like stem cell transplantation, enzyme replacement infusions, and physical therapy.

MPS1 is caused by an enzyme deficiency, and the initial screening test is looking for absent or low levels of this enzyme in the newborn's blood sample. Some unaffected infants may also express low levels of this enzyme, so follow-up testing is required. The current method of confirming whether or not an infant is affected is by sequencing the *IDUA* gene that causes MPSI.

The expansion in newborn screening for MPS1 means that GGC's diagnostic lab, the follow-up lab for SC's newborn screening program, is seeing more newborn screening samples coming through for confirmatory testing through gene sequencing.

"One significant complication that arises from this influx of samples is the identification of a large number of unique genetic variants in the *IDUA* gene," said Rich Steet, PhD, GGC's Director of Research. "Some of these variants are true mutations and the answer is clear, but often we are finding novel changes in this gene that are much more difficult to interpret."

“Some of these novel changes may be disease-causing, while others are not,” added Steet. "And because symptoms of MPS1 are not present at birth, not knowing the significance of the gene variants puts patients and families in a state of limbo, uncertain as to whether they should start therapy.”

Laura Pollard, PhD, Director of GGC's Biochemical Genetics Lab agrees, "The *IDUA* gene is also known for having what are called pseudodeficiency alleles, or genetic changes that mimic true mutations, but do not cause disease," she said. "It's so important that we distinguish between babies who are truly positive and need early intervention and treatment and those for whom invasive and expensive treatments are unnecessary."

Through the MPS Society funding and a grant from the National Institute of General Medical Sciences, GGC's research and diagnostic labs have developed a biochemical platform that helps distinguish which MPS1 variants are benign and which are disease-causing. Such "functional studies", which are designed to determine the impact, if any, of the new variant, are

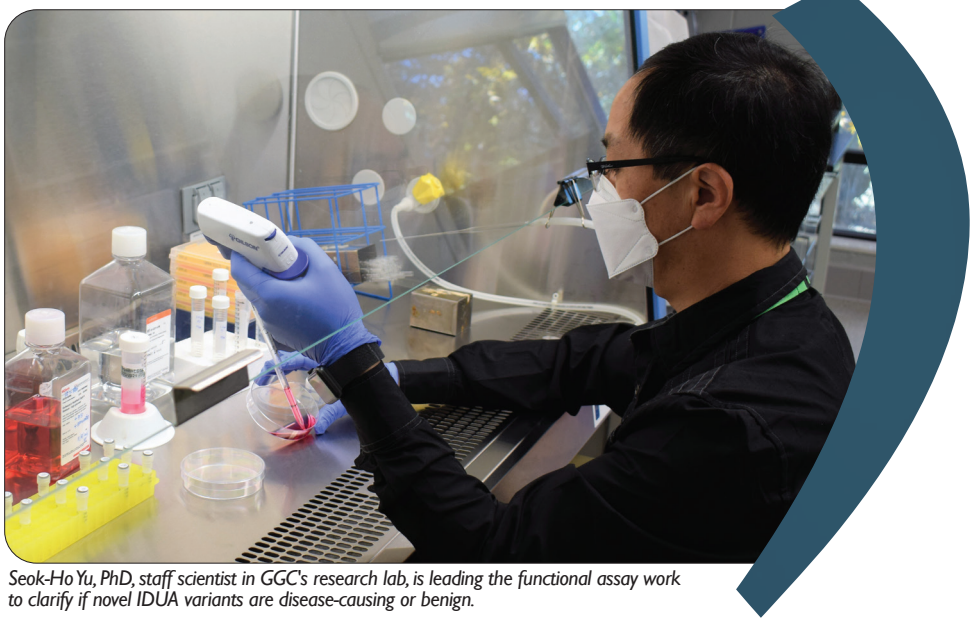
often the only way to assess if it is truly disease-causing.

"Most functional studies are long-term projects that can take months or years to clarify, and with worried parents and early treatments at stake, we don't have that kind of time," said Heather Flanagan-Steet, PhD, GGC's Director of Functional Studies.

"When we are uncertain of the meaning of a variant, we now have a platform to pursue functional studies more quickly to see how the variant is or isn't affecting the enzyme function,"

The functional studies platform was developed through a collaborative effort between Steet, Flanagan-Steet, and Pollard's laboratories. For patients with uncertain genetic variants, the laboratory uses an assay to more precisely measure the specific activity of the IDUA enzyme. This approach allows the lab to determine to what degree an uncertain variant is actually impacting the enzyme activity and therefore, whether it is predicted to cause disease.

"We have found that this new assay can distinguish between variants that are disease causing, those that are 'pseudodeficiency' alleles, and those that are benign," said Pollard. "Now, instead of sending back a report that basically says 'Your new baby may or may not be at-risk for a life-limiting disease,' we can take those extra steps and provide a clear answer." Since SC started screening for MPSI in February of 2021, GGC has received 65 samples for IDUA gene sequencing from babies who initially screened positive for MPS I. Of those, 14 infants were found to have a variant of uncertain significance, for which this new functional studies platform could prove beneficial.



Seok-Ho Yu, PhD, staff scientist in GGC's research lab, is leading the functional assay work to clarify if novel IDUA variants are disease-causing or benign.

The new platform is also having an impact beyond South Carolina.

"In addition to our work with the SC Newborn Screening Program, GGC also receives follow-up testing for infants in other states who initially screen positive for MPS1," shared Pollard. "We have heard from physicians outside of SC who are excited about our ability to perform these functional assays on variants identified in their patients."

The functional work on IDUA variants is also being expanded and adapted to be used for variants in other genes that cause different types of lysosomal storage disorders.

The biochemical platform was published in the *International Journal of Neonatal Screening* in late 2020.

New GGC Foundation Leadership

Cady Nell Keener, CFRE, will be assuming the role of Executive Director of the GGC Foundation in January.

Keener, a native of Abbeville, SC, is joining GGC after nine years working in development at Charleston Southern University (CSU) in Charleston, SC, most recently as Associate Vice President for Development and Special Gifts. She is a graduate of the University of South Carolina Darla Moore School of Business and earned an MBA from CSU.

"I'm grateful and thrilled for the opportunity to join the GGC family," said Keener. "As a native of the area who has been away for 15 years, it's exciting to return home and help impact my community. The GGC has many moving parts, and each one of the individuals on the team plays a significant role in impacting the lives of the families they serve. I am eager to begin meeting more teammates, patients, donors, investors, partners, and friends of the GGC to hear their GGC stories and explore ways to advance the mission."

"Cady Nell brings experience and enthusiasm to the GGC Foundation, and we are thrilled to welcome her into this leadership role," said Boo Ramage, Interim Executive Director of the Foundation. "She is already developing a plan to hit the ground running, and we look forward to adding her to our dedicated team."

TRAINING PROGRAM GRADUATES THREE

Kameryn Butler, PhD, completed a three-year Laboratory Genetics and Genomics fellowship, and **William Burns, MD,** and **Catherine Ziats, MD**, each completed a two-year residency program in Medical Genetics and Genomics through the GGC'S Medical Genetics Training Program in 2021.

Dr. Butler competed her undergraduate degree in Genetics at Clemson University and earned a PhD in Genetics and Molecular Biology from Emory University. At GGC, she studied the implementation and interpretation of molecular and cytogenetic diagnostic tests, as well as the development of new genetic testing assays for use in the clinical setting. Upon completion of the fellowship, Butler accepted a faculty position as Assistant Director in GGC's Diagnostic Laboratories working in both the Cytogenetics and Molecular Genetics labs.

Dr. Burns is a graduate of Tulane University and completed his medical degree at the University of South Carolina School of Medicine in Columbia. He completed a Pediatrics residency through Prisma Health-Midlands/University of South Carolina School of Medicine. Dr. Burns is now pursuing a fellowship in Medical Biochemical Genetics at Nationwide Children's Hospital in Columbus, Ohio.

Dr. Ziats is a graduate of the University of Florida and completed her medical degree at the University of Florida College of Medicine in Gainesville. Before joining GGC's program, she completed two years of training in Neurological Surgery at the University of Michigan. Dr. Ziats has joined the faculty at Dell Children's Medical Group in Austin, Texas as a Clinical Geneticist.

All three graduates earned their board certification through the American Board of Medical Genetics and Genomics this fall, Burns and Ziats in Clinical Genetics and Genomics and Butler in Laboratory Genetics and Genomics.

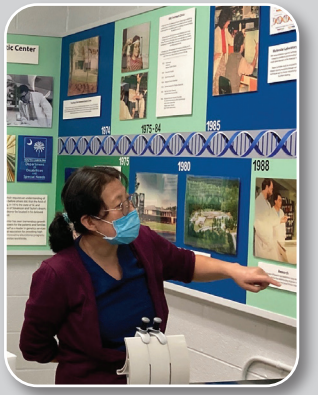
GGC's training program includes intensive education in laboratory technologies, clinical genetics, and diagnostic laboratory management. Since the training program began in 1989, 41 fellows and residents have completed the training and have gone on to practice clinical genetics or take leadership roles in genetics laboratories.

Dr. Kameryn Butler

Dr. Will Burns

Dr. Catherine Ziats

GENE WEEK - REVAMPED



Molecular Specialist, Xuemei Shi, MD, PhD, reads about GGC's history at the museum opening.

With COVID cases rising in early fall, GGC's annual Gene Week celebration was scaled back a bit this year.

GGC employees were treated to a socially distanced 'Grand Opening' of the GGC museum in the Genetic Education Center on September 28. Then, on October 1, GGC employees and their families, along with a few patient families enjoyed recognition as the 'Nonprofit of the Game' at the Greenville Triumph Soccer match.

To wrap up the week, we held a virtual Race the Helix-Greenwood on October 2, with great success! The event drew record-setting support from individual and business sponsors, both locally and around the globe. Approximately 200 individuals signed up to run or walk, many of whom shared their times and photos on social media to promote their participation. While we missed the in-person event, we enjoyed seeing how our supporters celebrated their own Race the Helix from Vermont to California, from mountain hikes to beach runs.

The Brown and Cameron families are ready to Walk the Helix!

Apprenticeships Inspire Future Scientists

The Greenwood Genetic Center has always placed great importance on education and inspiring students to enter a life science field. A long-standing partnership with the G. Frank Russell Technology Center in Greenwood has provided many interested high school students with unique work-based opportunities to explore their interest in the field of genetics.

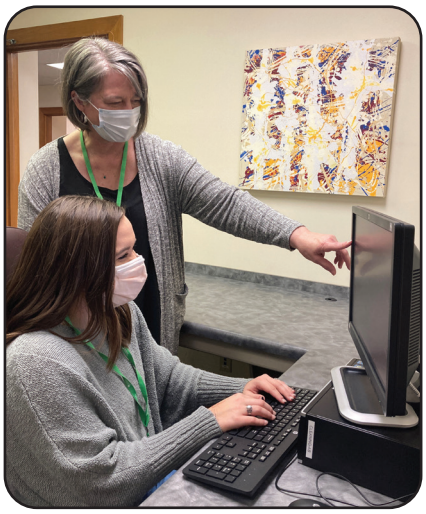
The apprenticeship program allows two high school students to become part-time employees for the school year. This year, Maggie Jones, a senior at Emerald High School, and Annabel Budreau, a senior at Greenwood High School, are each spending 15 hours per week working at GGC.

Jones, who aspires to earn a PhD and become a genetic researcher, works in the research division, assisting with zebrafish care and organizing DNA samples in the cold room.

"I enjoy so many things about working here but most of all I love the people," said Jones. "I have seen and learned so much and that is because everyone I meet here always tries to answer my questions, and if they cannot, they find someone who can or research it themselves."

Budreau, who plans to study biology in college and perhaps attend medical school, agrees, "The thing I enjoy most about my job at GGC are my coworkers. I have built relationships with multiple people in each facility and it allows me to gain a greater insight on what they do at the center."

Budreau splits her time at GGC between the administration offices and the cytogenetics lab experiencing both the business side of healthcare as well as the scientific opportunities.



"The Genetic Center has been a valuable partner to the Youth Apprenticeship Program for many years providing opportunities for our students in the Biomedical Sciences program to receive real world experience in the world of genetics and research," said Chip Whitt, Work-Based Learning Coordinator for the G. Frank Russell Technology Center. "Former students have used this program as a springboard to the Genetics program at Clemson University as well as other fields in the medical profession."

Above: Annabel Budreau works with HR Director, Janet Still, in digitizing GGC's administrative records.

Below: Maggie Jones feeds lunch to the 8,000 zebrafish in GGC's Allin Aquaculture Facility

Career Milestones

Recognizing employee achievements

The Greenwood Genetic Center is proud to employ some of the most dedicated, innovative, and talented professionals who always put our patients first.

We are thrilled to share their professional achievements.

Fatima Abidi, PhD, was promoted to Associate Director of the Molecular Diagnostic Laboratory.

Fran Annese has been named as GGC's Compliance Officer.

Brandi Buff was promoted to Chief Financial Officer.

Kameryn Butler, PhD, has been named Assistant Director in GGC's Cytogenetics and Molecular Diagnostic Laboratories. Dr. Butler also became certified in Laboratory Genetics and Genomics by the American Board of Medical Genetics and Genomics.

Jessica Cooley Coleman of the Molecular Diagnostic Lab was promoted to Laboratory Technologist, Level 4.

Brittany Hennigan of the Molecular Diagnostic Lab was promoted to Laboratory Technologist, Level 3.

Jill Johnson was promoted to Supervisor of the Cytogenetics Laboratory.

Ray Louie, PhD, was promoted to Associate Director of the Molecular Diagnostic Laboratory.

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

Janet Still was promoted to Director of Human Resources.

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.

Greenwood Genetic Center

106 Gregor Mendel Circle
Greenwood, SC 29646

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US POSTAGE
PAID
GREENWOOD, SC
PERMIT #225

106 Gregor Mendel Circle
Greenwood, SC 29646

Tel: 864-941-8100
Toll Free: 888-442-4363

www.GGC.org