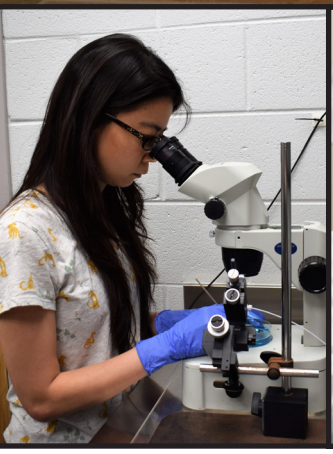
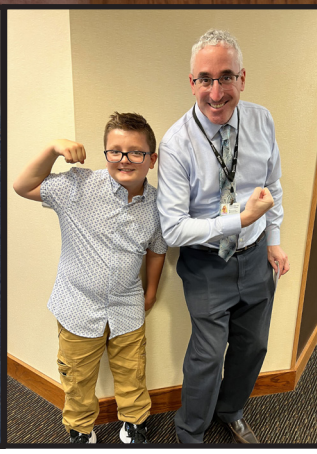
Greenwood Genetic Center

Where Compassion Inspires Progress





ANNUAL REPORT
2021-22

Dear GGC Friends,

We are excited to share this annual report with you - our wonderful GGC donors, friends, and supporters! The past year has been an exciting one, and we are grateful for your continued support allowing us to provide the highest quality care to the patients and families we serve.

As many of you are aware, GGC and the Medical University of South Carolina (MUSC) entered into a membership agreement earlier this year that joins the expertise of our two organizations under our similar missions. Our shared goal of improving access to care for all citizens of our state is driving this agreement, and we are already moving forward on joint initiatives through several working groups in areas including clinical care, technology, and education, finding wonderful cooperation among GGC faculty and our MUSC counterparts. Our new Board of Directors, which includes members appointed by both GGC and MUSC, are committed to and focused on advancing the mission and securing the ongoing success of the Greenwood Genetic Center.

GGC has also made great strides in technology over the past year. Last fall our diagnostic laboratories began offering whole genome sequencing to GGC patients, and we are already seeing more families receive that all-important diagnosis at an earlier point in their journey. Our researchers are advancing the knowledge and treatment possibilities for several rare diseases. Our clinical team is perfecting the use of asynchronous eVisits for patients, helping us significantly decrease wait times in our clinics and provide care and support that is convenient for our patients. Our education team is back to pre-pandemic numbers of outreach trips for middle and high schoolers, and they are developing genetics curricula for several healthcare provider programs across SC. Our business office and support staff are ensuring the Center's financial and operational stability and are supporting the efforts of all of our divisions. I'm so proud to be part of this excellent and collaborative team at GGC!

Thanks to your generosity, GGC continues to grow and thrive. In the following pages, you will find numbers that reflect our recent budget year and many of our activities across all divisions. You will also find stories of how your support is touching families directly. Your gifts are funding testing for patients who may not otherwise be able to afford it, providing innovative educational experiences for future scientists, and moving research breakthroughs closer to treatments.

On behalf of GGC's faculty, staff, and board of directors, I thank you for your continued support and commitment to our mission. We are deeply grateful that you have found our work - our passion - worthy of your support. We never forget that you are a most important member of the GGC family.

With deepest gratitude,

Steve Skinner, MD
GGC Director

On the Cover

1. Kathie Marsh, Dr. Julius Leary, and Lynne Lovett with a photo of their sister, Dianne, at the Leary Clinic dedication.
2. Jen-Jie Lee, PhD, works with zebrafish embryos.
3. Matt Tedder, PhD, leads efforts with methylation testing.
4. Physician assistant, Laura Buch, MSPAS, PA-C, counsels a patient.
5. Sophia Shockley works with fruit flies in GGC's summer camp.
6. Rich Steet, PhD, flexes with Brody Baker at the Leary Clinic dedication.

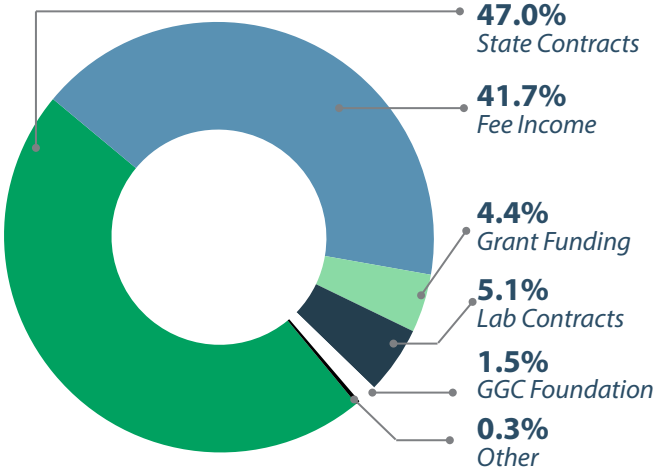
Our Impact

The Greenwood Genetic Center is committed to offering compassionate care to all families across the lifespan who are impacted by genetic disorders. Through advanced technologies and a grant from The Duke Endowment, GGC is improving access to clinical services using technology and asynchronous care to remove barriers and reduce wait times for appointments. The GGC Foundation's GGC Cares Fund is providing testing to many patients who would have otherwise been unable to afford these necessary services. Your gifts to the GGC Foundation have allowed us to continue to provide genetics care and hope to families across our state and around the world when they needed it most.

In 2021-22, your generosity helped us to:

- Complete **5,208** clinical patient appointments (**2,870** in person, **2,219** through telegenetics and **119** through eVisits);
- Report **20,723** laboratory tests to find long sought-after answers for patients;
- Prevent **70** neural tube defects through the SC Birth Defects Prevention Program;
- Coauthor and publish **52** scientific manuscripts and **2** book chapters to share our advances and expertise with the broader medical community;
- Inspire **8,690** students through **151** mobile lab visits and field trips;
- Initiate **25** functional research studies in cells and zebrafish to clarify uncertain genetic testing results. This led to **9** definitive answers and **2** new targets for treatment; and
- Bring in **$1.1M** in grant funding through **3** clinical projects, **3** NIH grants and **3** private foundation grants.

GGC Revenue Sources



Income ($24.2M)

Our Mission

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

Our Vision

The Greenwood Genetic Center will be a Center of Excellence in Medical Genetics, serving as a resource for all persons who need genetic services or information, and working to reduce the prevalence and impact of genetic disorders.

Annual statistics reflect activities from July 1, 2021-June 30, 2022, GGC's most recently completed fiscal year. The financial data are unaudited as of the publication date.

From One Family to Another

Dr. Julius Leary meets Brody Baker who has benefited from his family's GGC gift.

A gift to the GGC Foundation in memory of a sister provides answers and hope to a family 900 miles away.

Nine-year-old Brody Baker of Atlanta, Missouri, jumped into a swimming pool before it was completely filled. "Not a great idea," recalled his dad, Aaron, and dad was right. Brody broke his tibia and ended up in a cast. But after the cast came off, even with physical therapy, he was left with a limp. Something else was wrong.

After visits to several specialists and numerous imaging studies, blood draws, and other tests, an astute orthopedist noticed Brody's highly arched foot, a deformity known as pes cavus.

"That's the first time we had ever heard of Charcot-Marie-Tooth disease (CMT)," said Aaron. "He told us not to Google it, that it would just be scary and to let them continue to do some testing." But Google it they did. "For me, I like to understand the science of it," said Aaron.

Genetic testing revealed an alteration in Brody's AIFM1 gene - a gene that has been linked to CMTX4 otherwise known as Cowchock Syndrome, but, as is common for rare diseases, Brody's was a unique change that had only been reported once before. The doctors, including a leading expert on CMT, Dr. Michael Shy, who practices just three hours from the family's home at the University of Iowa, suspected that the genetic change was significant, but needed more proof.

After the suspected diagnosis of CMTX4 was made, Google is also what led the Baker family to the Greenwood Genetic Center (GGC). "I found an article online about GGC's research into CMT that was made possible by a memorial fund honoring Dianne Leary, who had a different type of CMT than Brody," said Aaron who then reached out to GGC Director, Dr. Steve Skinner, to see if there was interest in Brody's variant and his rare type of CMT.

Dianne's siblings, Julius Leary, MD, Lynne Leary Lovett, and Kathie Leary Marsh, established the Dianne Patricia Leary Fund for Charcot-Marie-Tooth (CMT) Neuromuscular Disorders at GGC in 2020 with a gift to the GGC Foundation in memory of their sister who passed away in April of that year. The gift is being used to further research into neuromuscular disorders like CMT.

Dianne Patricia Leary

Dr. Rich Steet, GGC's Director of Research, spoke with Dr. Shy and University of Iowa genetic counselor, Tiffany Grider, and arranged for fibroblasts (skin cells) from Brody to be sent to GGC. "In collaboration with Duke, we had already studied another variant in this gene and had all the methods in place to study Brody's variant. Our work showed that his variant impacted the AIFM1 protein in a very similar way to the prior patients, meaning that it is causative for CMTX4," said Steet.

The diagnosis was now clear, but what does it mean? What's next?

Brody's CMT course thus far has been mild. "He does fall down some," said his dad, but he shows no signs of the cognitive impairment that is often seen in CMTX4.

The Baker family has built a virtual CMTX4 community, albeit a small one because of the rarity of this diagnosis. Through a CMT Facebook group, they have connected with a few other CMTX4 families including an individual who is deaf, but owns a restaurant and has no cognitive challenges, as well as a family with two cousins who both have developmental issues.

"Even though Brody's variant affects the AIFM1 protein in a similar manner to the other CMT4X patients we studied, his clinical features appear milder," added Steet. "We are working with the Baker family, Brody's medical team, and other collaborators to develop new cell and animal models for this condition that can be used to understand why there is such variability between patients, and ultimately find therapies for CMTX4."

Brody Baker with one of his chickens at a 4H show

"Because of how rare the disease is, Dr. Steet's approach – how he's hoping to do the research - will be very helpful because we just don't have enough patients in the US," said Baker. "This research is a step in the right direction, and it will be some of the only research on CMTX4."

Even with all of the doctor visits and tests, Brody remained confused about his ongoing limp. "He didn't understand that it wasn't a remnant of his broken leg, so to help him better understand this diagnosis, we asked him to do a presentation for his 4-H club about CMT," said Baker. "That project not only helped him to better understand the diagnosis, but also helped him process his feelings about it."

Brody is determined to not let this diagnosis slow him down. He plays basketball and shows chickens and 1,000-pound steer through his 4-H program in Missouri. In addition to talking about his diagnosis at 4-H, he also has a YouTube channel to help others better understand what CMT is and how it impacts his day-to-day life.

In October, GGC officially dedicated 'The Leary Clinic' in Greenwood in memory of Dianne. Aaron and Brody Baker flew in from Missouri for the event, and Brody, now 12, spoke to the audience, "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."

"Our family is hopeful for Brody's future and grateful to GGC and the Leary family for providing resources to advance the understanding of this rare group of disorders," added Baker.

"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."

More about CMTX4

CMT diseases are a category of inherited disorders that cause peripheral neuropathy, or nerve damage in the extremities, that leads to muscle weakness, loss of sensation, and difficulty walking. Foot deformities are common. Other features that can be present in patients with CMTX4 include intellectual disability and hearing loss, There are over 100 genes that have been found to cause the various forms of CMT.

Brody's specific type of CMT, CMTX4, is X-linked, meaning that the gene that causes it, *AIFM1*, is located on the X chromosome. Males have a single X chromosome, so when that gene is altered in a male, they will express the disease. Because females have two X chromosomes, those who have an *AIFM1* mutation are not affected, but can pass it on to their sons.

Running with Compassion!

Race the Helix events returned in person to support the GGC Cares Fund

GGC's very dedicated employees, supporters, volunteers, and sponsors were thrilled to be back in person for the 7th annual Race the Helix - Upstate in Greenville, SC in April and 12th annual Race the Helix - Greenwood in October! Hundreds of walkers and runners joined the in-person events with others participating virtually. One of the event's presenting sponsors, Bionano, even held their own Race the Helix for employees in San Diego with many of their remote employees participating virtually from all over the US!

Proceeds from Race the Helix are directly benefiting families through the GGC Foundation's Cares Fund. In line with the Center's focus on improving access to genetics care and services, the GGC Cares Fund helps to remove some of the financial burden from qualifying families who are uninsured or under insured. To date the GGC Cares Fund has covered nearly $25,000 in testing needs for GGC patients. Donors can contribute directly to the GGC Cares Fund by visiting ggc.org/foundation/donate.

Right: JT Shorter, a GGC patient, won 'Race the Helix' Greenwood. Left: Employees from Bionano at Race the Helix-San Diego!

Thank you!

On behalf of GGC's faculty and staff, our patients and families, and all who are served by GGC, we express our deepest gratitude for these individuals, families, and organizations who support the Center's life-changing work.

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