Greenwood Genetic Center

Summer 2019

A Newsletter for the Friends of the Center

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GGC Co-Founder Roger Stevenson, MD Honored with ACMG's Highest Award

GGC co-founder, senior clinical geneticist, and Ravenel Boykin Curry Chair in Genetic Therapeutics, Roger E. Stevenson, MD, was recognized with The American College of Medical Genetics and Genomics Foundation's David L. Rimoin Lifetime Achievement Award in Medical Genetics. The award was announced at the College's annual meeting in Seattle on April 2.

Stevenson's genetics career took hold early when, as a medical student at Bowman Gray School of Medicine at Wake Forest, he set up the school's first chromosome lab and made a groundbreaking discovery about the risk of birth defects in mothers with PKU, a rare inherited metabolic disorder. He proceeded to Johns Hopkins University School of Medicine where he completed a pediatric residency and research fellowship. It was there he studied under some of the giants of the emerging field of medical genetics and was a contemporary of the award's namesake, Dr. David Rimoin.

While studying at Johns Hopkins, Stevenson met another research fellow, Harold Taylor, PhD, with whom he formed the Greenwood Genetic Center in 1974 with support from Greenwood businessman and philanthropist Jim Self and the SC Department of Disabilities and Special Needs (DDSN). Since that time Stevenson has led the Center to international acclaim.

With special interests in intellectual disability, birth defects, and autism, Stevenson has published over 200 scientific papers, and numerous widely utilized texts and reference books.

In 1978, he identified a rare genetic disorder that bears his name, Beare-Stevenson syndrome, and several other disorders that, because of his tendency to avoid personal recognition, do not.

Over the past twenty years, he and GGC research colleagues, including Dr. Charles Schwartz (p. 8) have identified nearly 1/3 of all known genes on the X chromosome that lead to intellectual disability, making GGC the international authority in this area of medical genetics.

One of Stevenson's great professional passions lies in the area of birth defects prevention. In 1992, in collaboration with SC DDSN, Department of Health and Environmental Control, and the March of Dimes, he founded the SC Birth Defects Prevention Program at GGC which has led to a 60% decline in severe defects of the brain and spine in SC. He champions efforts to educate the population on the importance of folic acid as a preventative supplement and led the charge to fortify dietary staples such as breads, pasta, and cereals with folic acid to help prevent these birth defects. This program has been hailed as a model for other states, and even other nations, by the Centers for Disease Control and Prevention.

Stevenson also holds a talent for teaching. GGC's Medical Genetics Training Program has educated dozens of MD and PhD geneticists who count him as their mentor.

Despite his many national and international accolades and prominence in the field, Stevenson continues to prioritize patient care. He is highly regarded by his patients and their families, not because of awards or publications, but because of his gentle nature and genuine compassion and concern for those he serves.

After relinquishing role of GGC Director in 2011, Stevenson continued an active clinical practice and is very involved in research, particularly as it relates to treatment of genetic disorders. In his role as Curry Chair, he oversees the Center's treatment initiatives including clinical trial participation and the adoption and delivery of novel therapies.

The Rimoin Lifetime Achievement Award recognizes an individual in the medical genetics profession who exemplifies a lifetime of achievements including a passion for teaching and mentoring, care of patients and their families, and enthusiasm for integrating the medical genetics and genomics community into mainstream healthcare.

IN THE WORDS OF HIS NOMINATORS

"Roger could have been chair of any department of genetics in the country and would likely have been a medical school dean had he chosen that path, but he chose an important different track to the benefit of children and families worldwide."

-Godfrey Oakley, MD, medical school colleague and Director of the Center for Spina Bifida Prevention at Emory's Rollins School of Public Health

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"Roger has an instinctive manner with families that puts them at ease and lets them know that he views their child as the most important person in the room."

-Katy Phelan, PhD, trainee and former GGC laboratory director

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"Roger has always been about ten years ahead of everyone in seeing what is important, what the new developments are, and putting them into practice,"

-Judith Hall, MD, Stevenson's colleague at Johns Hopkins, longtime friend and collaborator, and 2018 Rimoin honoree

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"Beyond the respect I have for Roger as a clinician, scientist, and educator, I have even greater regard for him as a man. While accolades and achievements can often lead to arrogance and pride – Roger embodies the opposite traits. He sees his work as that of a servant providing care and hope to families struggling with difficult circumstances."

-Steve Skinner, MD, trainee and current GGC Director

"From the moment I met Roger, he has never been anything except kind, forward-thinking, compassionate, and genuinely interested in impacting the world around him in the smallest or largest of ways, with the utmost humility every step of the way."

-David Everman, MD, GGC clinical geneticist

SHARING AUTISM EXPERTISE

With over 20 years of research in the study of autism spectrum disorder (ASD), GGC has amassed a great deal of data and expertise in this increasingly common disorder. Dr. Luigi Boccuto, Assistant Research Scientist, leads the Center's research efforts in developing a blood based test for autism. He also serves as the Chief Medical Officer for Swiss company, STALICLA, where he is assisting with the development of treatments for subgroups of patients with ASD. This spring, Boccuto was invited to share his work at two Upstate events.

At the Converge Autism Summit, a symposium for educators, parents, and professionals, he spoke on 'Genetics and Environmental Aspects of Autism Spectrum Disorder: New Models and New Theories' (pictured left).

GGC also cohosted the 3rd annual Human Genetics Symposium with Bob Jones University where he shared 'Personalized Medicine in Autism: New Approaches to an Old Problem.' At this sold out event, GGC's Carrie Buchanan, MD also presented "Autism Spectrum Disorder: A Clinical Perspective in Pediatrics,' and Jennifer Stallworth, MS, CGC lead a workshop on genetic counseling for families impacted by ASD.

SC LT. GOVERNOR VISITS GGC

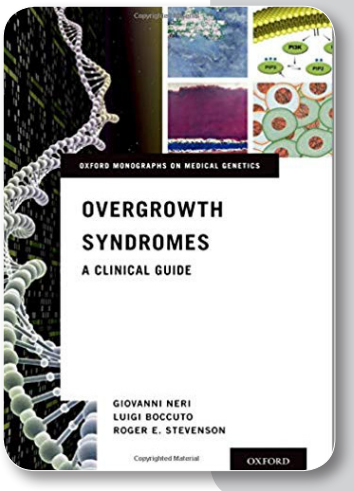
South Carolina Lieutenant Governor, Pamela Evette, visited the Greenwood Genetic Center's Greenwood campus in April to learn more about what the Center is doing for families across the state. She met with GGC leaders, as well as the Greenwood Partnership Alliance and local legislative delegation, and toured GGC and the Clemson Center for Human Genetics.

Lt. Gov. Evette was informed about the Center's long history in serving families with a variety of genetic disorders, as well as the financial impact of GGC on the state. GGC Director Steve Skinner, MD, shared that GGC's prevention and treatment programs save the state over $85 million per year in health care costs, compared to the state's investment in the Center of just over $4.5 million annually.

Heather Simmons Jones of the Greenwood Partnership Alliance also shared the strategic and marketing plans for the Greenwood Genetic Center Partnership Campus which aims to attract academic, industrial, and medical organizations to the Center's campus.

Evette shared her enthusiasm for the work being done at GGC and applauded the collaborative spirit of the faculty. "My goal is to make connections and facilitate collaborations," she said. "I can use what I learned here about the great work you are doing and share that, both in Columbia and in my travels all over the state."

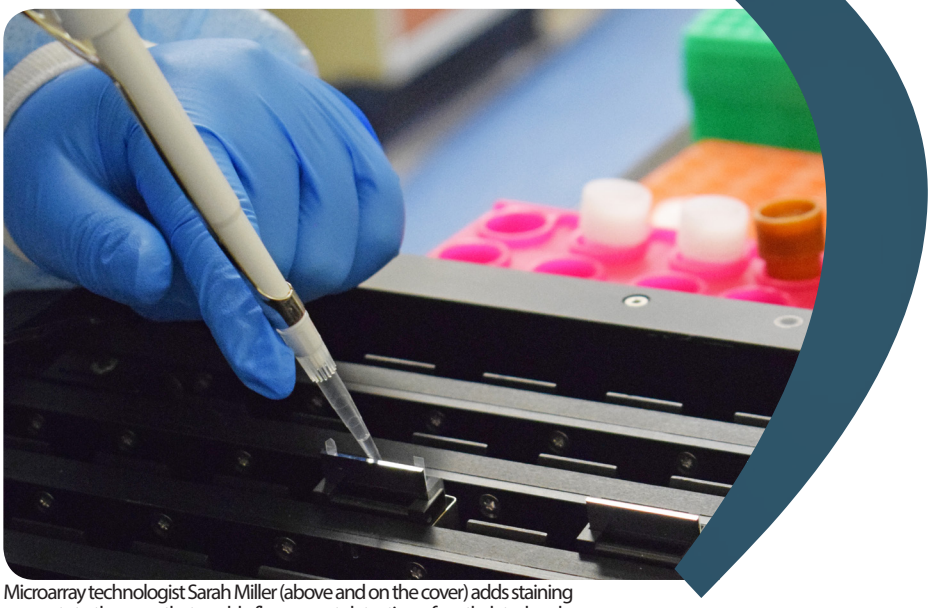
GGC FACULTY AUTHOR NEW REFERENCE



Giovanni Neri, MD, retired Professor of Medical Genetics at Catholic University in Rome and GGC Senior Genetics Scholar is an international expert on genetic disorders involving overgrowth. Neri, along with his former student and current GGC Assistant Research Scientist, Luigi Boccuto, MD, and GGC Senior Clinical Geneticist, Roger Stevenson, MD, recently edited Overgrowth Syndromes: A Clinical Guide, part of the Oxford Monographs on Medical Genetics series. The text was published in March.

Overgrowth syndromes are rare, with a combined incidence of just over 1 in 10,000 and include disorders such as Beckwith-Wiedemann syndrome, Sotos syndrome, and Proteus syndrome. Manifestations of these conditions often extend beyond body size and may involve other concerns such as intellectual disability, metabolic abnormalities, or increased risk of malignancy.

This book is a comprehensive guide to genetic disorders that cause overgrowth, both generalized overgrowth which affects the entire body, or segmental overgrowth where only one tissue or body part grows excessively. With information on both the clinical features of these syndromes as well as laboratory diagnoses, clinicians can better understand these disorders and the genes that are known to cause them.



Microarray technologist Sarah Miller (above and on the cover) adds staining reagents to the array that enable fluorescent detection of methylated and unmethylated areas of the DNA.

EPISIGN

GGC FIRST LAB IN THE WORLD TO OFFER NEW DIAGNOSTIC TEST

Greenwood Diagnostic Laboratories at GGC, in partnership with London Health Sciences Centre (LHSC) in Canada, launched a new diagnostic test focused on disease-specific epigenetic signatures. The test, EpiSign, analyzes markers of DNA expression rather than sequence changes to establish a diagnosis or help resolve variants found through DNA testing.

GGC's Dr. Charles Schwartz, Senior Research Scientist, worked closely with Dr. Bekim Sadikovich at LHSC to assemble cohorts of patients with potentially unique methylation signatures.

GGC was the first in the world and is currently the only US laboratory to offer this new technology clinically.

"Almost all genetic testing is focused on looking at the DNA code to identify mutations or changes to that code that can lead to disease," said Mike Friez, PhD, Director of GGC's Greenwood Diagnostic Laboratories. "Epigenetics is a newer concept looking at changes in gene expression. So even though the genetic code may be normal, if a gene is over- or under-expressed, it can lead to a genetic disorder."

Genes can be turned on or off through the process of methylation, or the addition of a chemical tag to a regulatory area of the gene. EpiSign analyzes patient blood samples for a specific pattern of methylation. To date, specific patterns, or epigenetic signatures, have been identified for 19 genetic disorders, many of which have overlapping clinical features making the diagnosis difficult.

The test can currently identify disorders including CHARGE syndrome, Cornelia de Lange syndrome, Down syndrome, Kabuki syndrome, Sotos syndrome, and Williams syndrome. EpiSign data is also useful for patients with a suspected diagnoses of disorders that involve abnormal DNA methylation such as Fragile X syndrome, Prader-Willi syndrome, Angelman syndrome, and Beckwith-Wiedemann syndrome.

Additional disorders are under review, and the panel of conditions detected by EpiSign is expected to grow rapidly.

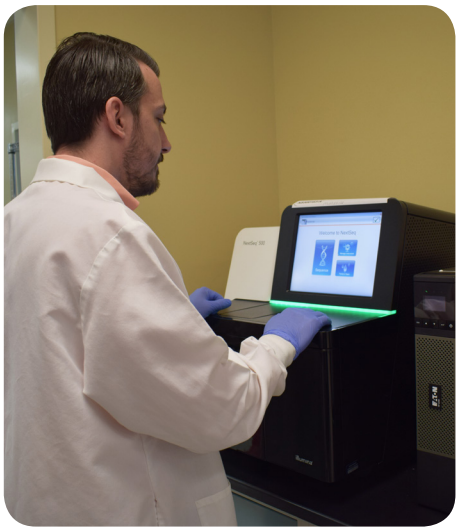
The test may also provide useful information for another group of patients, those in whom a variant has been found from DNA sequencing.

RECORD-SETTING YEAR

GGC's Biochemical Genetics Laboratory posted a record-setting year in 2018. The lab finished the year with 10,703 signed-out laboratory tests, up from just over 10,000 in 2016 and 6,300 tests in 2015. Much of the increase can be attributed to several partnerships between the lab and numerous pharmaceutical companies. Through these collaborations, GGC is helping to identify patients, provide appropriate diagnostic tests, and monitor the effectiveness of several therapies that are part of ongoing or upcoming clinical trials.

"We are excited to be involved in work on several emerging treatments that have the potential to impact the quality of life for patients in significant ways," shared Tim Wood, PhD, Director of GGC's Biochemical Genetics Laboratory (pictured below, far right). "I can't say enough about the dedication of our lab team who puts in hard work day after day to make sure our patients receive the most accurate and timely test results."

GGC's Biochemical Genetics Laboratory faculty and staff



GGC staff scientist, Matt Tedder, PhD, sets up the EpiSign chip for analysis.

"When we perform gene sequencing, it is common for a patient to have what we call a variant of uncertain significance or VUS," shared Kellie Walden, MS, CGC, laboratory genetic counselor for Greenwood Diagnostic Labs. "A VUS result means that we are unsure if what we found is a disease-causing mutation or simply a normal variant. EpiSign will be able to help us better classify the VUSs in these genes as harmful or benign, giving the patient a clear answer and path forward for medical care."

Epigenetic variants with unclear impact/significance have also been found with increased frequency in patients with unexplained intellectual disability (ID) and/or congenital anomalies.

Friez noted that achieving an accurate diagnosis for conditions involving ID and/or congenital anomalies can be challenging, "These patients often have variable, complex and overlapping features. Even by using the most advanced technologies at our disposal, we are only able to identify a cause in 42-62% of patients, leaving many without a clear diagnosis. EpiSign is another tool now at the clinician's disposal to help end the diagnostic odyssey for many families."

GGC launched the new test during the American College of Medical Genetics and Genomics (ACMG) annual meeting in Seattle where Sadikovich shared the work that led to the test development in a platform presentation.

At the GGC booth at the ACMG meeting with an EpiSign 'Lite Brite' are (L-R) Walden, Friez, and marketing specialist, Caroline Pinson.

Using the tagline 'Methylation Understood', a light bulb theme at the GGC booth illustrated the new understanding of how methylation signatures can be used to diagnose patients complete with a giant EpiSign 'Lite Brite.'

EpiSign
methylation understood

WHAT IS EPIGENETICS?

Humans have approximately 20,000 genes, but all of those genes collectively make up only around 1-2% of our DNA. In recent years, we have learned that the other 98-99% is not 'junk DNA', but does seem to serve a vital purpose. Much of it controls the expression of genes.

Epi- means 'upon' or 'on', so epigenetics is the study of the DNA elements and other influences that have an impact upon the genes. These influences control how those 20,000 genes are expressed. They turn genes on, off, up, or down depending upon the cell type, the individual's stage of development, and the particular need for the protein encoded by that gene at that time.

Epigenetic influences are chemical alterations or tags that are added to the DNA, usually consisting of a methyl group (CH_3) attached to the DNA strand. Methylation can be triggered by any number of genetic and environmental factors. Age, sleep patterns, diet, and exercise habits can all cause methylation to occur which affects gene expression. Other such modifications are inherited.

GGC's EpiSign looks for alterations to the genes that do not involve changes to the gene sequence, so they aren't detected using traditional genetic sequencing technologies. Scientists have currently found 19 disorders that can be identified and distinguished from one another based on unique methylation signatures, and that list will grow quickly.

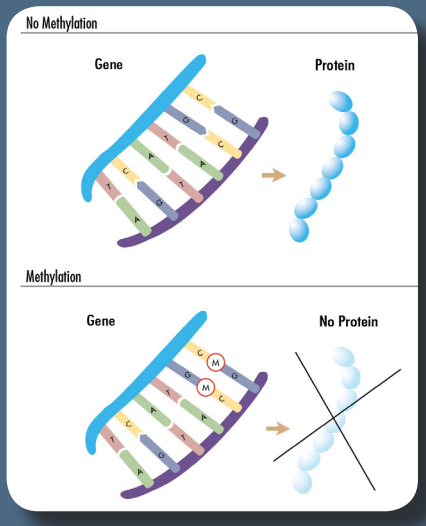
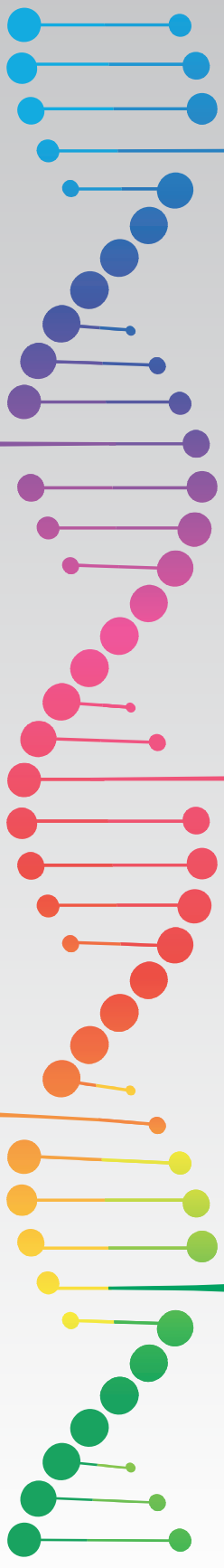


Figure: The DNA sequence of both genes is the same, but the addition of the methyl group (M) to the lower image prevents the gene from producing a protein. GGC Counseling Aids, 6th ed.

EDUCATIONAL IMPACT

After nearly a decade on the road, GGC's educational outreach programs continue to grow at a rapid pace, reaching teachers and students across SC providing engaging activities and encouraging career exploration.



200

Trips made by GGC's Outreach Education faculty and staff on the Gene Machine and Helix Express in the 2018-19 school year. This includes visits to 126 different middle and high schools, both public and private, across SC.

12,952

Students served by the Gene Machine Mobile Science Lab, Helix Express Van, and Genetic Education Center during the 2018-19 school year.

140,776

Miles travelled on the Gene Machine since it first hit the road in the fall of 2010.

42/46

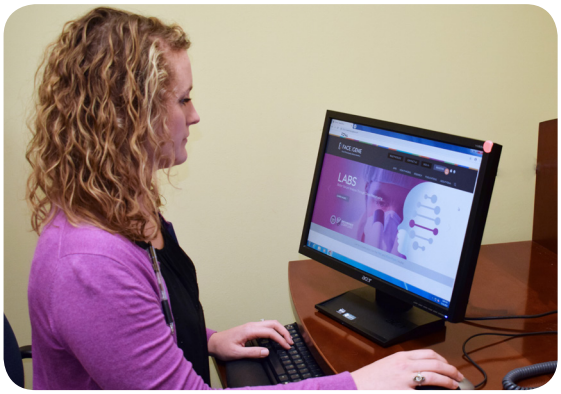
South Carolina counties served by GGC's Educational Outreach Programs since 2010.

>60,000

Students served by the Gene Machine Mobile Science Lab (2010-present), Helix Express Cargo Van (2017-present), and Genetic Education Center (2010-present).

Filling the 'GAPP'

Using technology to improve care



There is a nationwide shortage of clinical geneticists - just half of genetics residency spots are filled each year, and training programs struggle to keep up with the demand for genetic counselors. This shortage increases the workload for clinicians at GGC and lengthens the wait time for worried families to access the care they need.

"A wait time of several months is unfortunately not uncommon, not only at GGC, but for genetics clinics nationwide," said Mike Lyons, MD, Director of Clinical Services at GGC. "A major goal of our clinical division is to shorten the time to the diagnosis, which not only gets the patient to appropriate therapies sooner, but also decreases anxiety and frustration among families."

To meet this need GGC is engaging state-of-the-art technology to improve patient care. Through a very successful telegenetics program, which began over two years ago, the Center has improved access for patients in the Pee Dee region of SC, allowing patients to be evaluated by genetics providers at other locations. This program has reported high patient satisfaction.

Another initiative to harness the power of information technology is also underway at GGC. The GAPP (Genetics Access in Primary Pediatrics) project, is a pilot program that links GGC's clinical geneticists and genetic counselors to pediatricians through an app called Face2Gene.

Face2Gene, a product of Boston-based FDNA, analyzes patient photographs using machine learning and computer algorithms to help geneticists make challenging diagnoses.

Now, through the GAPP pilot project, the capabilities of Face2Gene are being used to improve patient wait times. GAPP allows a pediatrician to identify patients through a list of genetic 'triggers' or features that may indicate a need for further genetic evaluation.

If the patient's family elects to be a part of the GAPP pilot project, the pediatrician then uploads facial photos and other clinical information to Face2Gene where it can be securely shared with GGC clinicians for review. The geneticist can suggest appropriate referrals or genetic testing that can be initiated by the pediatrician in advance of the genetics appointment. Urgent referrals can be prioritized, and when the patient does come in for their genetics consultation, initial test results have already been completed, saving valuable time.

Deborah Greenhouse, MD, of Palmetto Pediatric and Adolescent Clinic in Columbia, one of a few SC pediatricians testing out the program, was excited to be a part of this initiative, "I honestly think that GAPP is one of the most innovative projects that I have had the opportunity to be involved with during my entire career, and definitely one of the most likely to have a very positive impact on patient care."

"Through the GAPP project, our hope is to improve the care our patients receive as well as make communication more streamlined between the referring providers and our clinics," said Hannah Moore, MS, CGC, lead genetic counselor on the project (pictured above). "We have great hope that GAPP will be beneficial for all parties involved and will be accessible to more providers in the near future."

A PAth for better Patient care

In addition to using new technologies to alleviate the shortage of medical geneticists, GGC is also involved in promoting careers in genetics within the physician assistant (PA) community.

GGC's first PA, Wesley Patterson, PA-C (pictured top), is actively recruiting PAs into careers in genetics. In 2018, he cofounded The Society of Physician Assistants in Genetics and Genomics (SPAGG), a professional organization comprised of PAs in the specialty of genetics. SPAGG is dedicated to the education, advocacy, and placement of PAs in genetics clinics in order to increase patient access to quality care. Patterson works closely with other GGC clinicians and cares for a wide variety of patients through clinics in GGC's Greenwood and Columbia offices.

GGC's Division of Education has also received a $82,000 grant from Sanofi Genzyme to expand PA education in genetics, especially as it relates to lysosomal storage disorders (LSDs), a group of rare conditions that are thought to be under diagnosed, but for which many medical management options exist. Dr. Leta Tribble, GGC's Director of Education, is leading efforts to provide lectures for PA students, create reference material specifically for PAs on these conditions, and plans to attend and present at national PA conferences. GGC is also developing materials for the website of the American Association of Physician Assistants where there are currently no genetics resources.

GGC welcomed its second PA this spring in the Greenville office. Laura Gardner, MSPAS, PA-C (pictured above) sees patients with genetic indications and is also participating in the Metabolic Advanced Practice Provider (MAPP) Fellowship, also funded by Sanofi Genzyme. MAPP is a 24 month training program designed to prepare a nurse practitioner or PA to assist in the diagnosis and medical management of individuals with LSDs.

Gardner is being mentored by Dr. Curtis Rogers in GGC's Greenville office, where the majority of LSD patients are followed.

A TIRELESS ADVOCATE

Schwartz, left, and Dr. Luigi Boccuto review plates from the team's autism experiments.

Charles Schwartz, PhD has been integral to the successes of the Greenwood Genetic Center for over 34 years. As he prepares to retire, it is fitting to recognize his tremendous contributions to GGC and the field of genomic medicine.

Charles E. Schwartz, a native of New York City, has dedicated his professional career to the diagnosis, understanding, and treatment of individuals with intellectual disability. He was recruited to GGC in 1985 from the University of Utah to start a molecular diagnostic laboratory, bringing a new level of technology to bear for families with intellectual disability and rare genetic disorders – families who had been struggling for answers, often for many years. Following the successful implementation of the molecular laboratory, Schwartz turned his focus to research in 1995, and was named Director of Research at GGC in 2004.

Through his experience, expertise, and commitment to serving patients and their families, he has built GGC's JC Self Research Institute for Human Genetics into a world class, internationally renowned research center with a focus on intellectual disability, birth defects, and autism. He was the first to attract NIH grants to GGC with over $12 million dollars in research funding over 25 years. He has contributed broadly to the literature, authoring 13 books/book chapters, and over 350 scientific publications. His influence in the field of medical genetics is immense.

Schwartz is recognized internationally as a leader in understanding X-linked intellectual disability (XLID, see right). Schwartz and his research lab in Greenwood have been involved in the identification of nearly 1/3 of the XLID genes, providing answers for families, helping to prevent recurrences of these disabilities, and moving the paths of treatments forward.

Schwartz's work has led to the launch of many clinical genetic tests that have provided countless patients with a diagnosis - a diagnosis that, because of the rarity of these syndromes had been difficult, if not impossible, to obtain prior to his discoveries. Most notably, he adapted the technique used to discover the gene for Fragile X syndrome, and GGC was the first lab in the US to offer clinical testing for this most common X-linked cause of intellectual disability.

His work in this field has led to the identification of the cause for many rare genetic syndromes including Renpenning syndrome, Snyder-Robinson syndrome, Christianson syndrome, Miles-Carpenter syndrome, and Allan-Herndon-Dudley syndrome.

"The tireless efforts of Dr. Charles Schwartz not only established GGC as an international hub for X-linked intellectual disabilities but drove the integration of basic research in genetics into the culture and mission of the Center," shared Rich Steet, PhD who has assumed the role of Director of Research. "His influence and leadership in both of these areas will be felt for years to come"

Dr. Schwartz has also worked to transform the way scientists and clinicians view autism spectrum disorder, a condition that impacts 1 in 59 children in the US. Since 1995, through the SC Autism Project, Schwartz's scholarship into the genetic causes of autism has led to the identification of several genes that are associated with ASD. His team's discoveries into the biochemistry of autism are contributing to the development of a blood-based test for ASD and are also informing the development of medical treatments and clinical trials.

Dr. Schwartz also has a long-standing interest in birth defects research, having identified several genes and chromosomal abnormalities that are responsible for defects of limb development.

ETHAN'S GARDEN

If you read Dr. Charles Schwartz's scholarly work, each of his scientific papers ends with a dedication:

In memory of
Ethan Francis Schwartz
1996–1998

The short, but influential life of Charles and Christine Schwartz's youngest son has served as inspiration and motivation for his tireless work on behalf of children and families worldwide. For many years, a statue of Ethan has graced his father's office. Now, upon his retirement, Ethan's statue will be placed in the center of the Greenwood campus in 'Ethan's Garden.'

The Garden will be dedicated in memory of Ethan and in honor of Charles's commitment to improving the lives of all children. Ethan's Garden and his likeness will inspire all of us at GGC - a daily reminder, as it has been to his father, of the precious gift of a child and the lasting impact they have on us all.

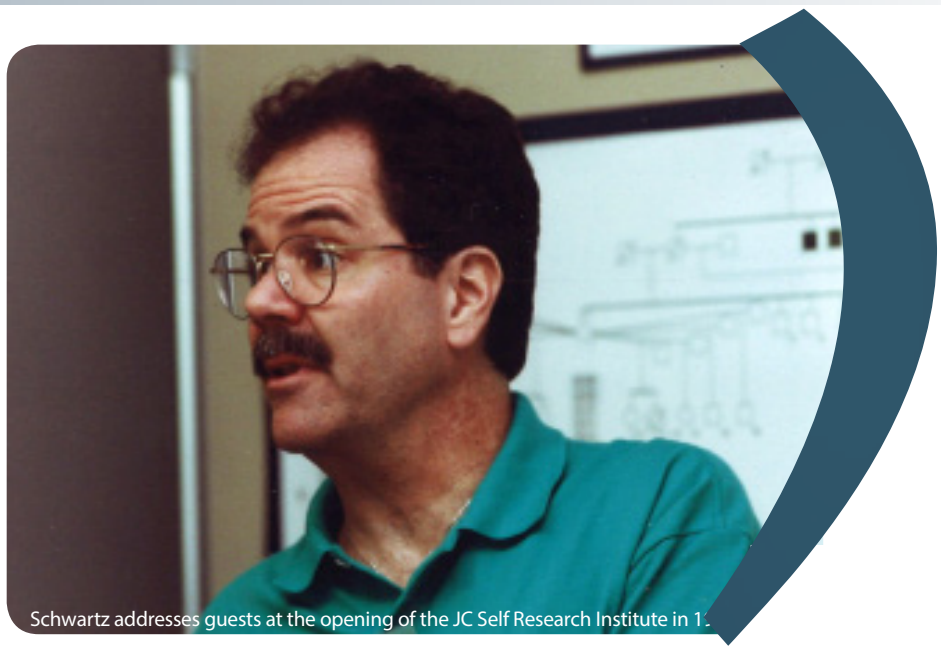
"Charles was and remains dedicated to the global mission of GGC," said Mike Friez, PhD, Director of GGC's Diagnostic Laboratories. "By working collaboratively and leveraging the resources of the Center for the betterment of those faced with a genetic disability he has helped to create a higher standard of patient care that GGC proudly carries forward."

In addition to his work ethic and expertise in these areas, Schwartz has a true understanding of the importance of his work on a personal level. He is involved with numerous family support organizations and foundations, and regularly attends their meetings not only to share research advances and answer questions, but also to spend time learning from the families that his work is impacting.

His work has been recognized with awards from the American Association on Intellectual and Developmental Disabilities (2002) and its SC chapter (2003). In 2015 he was honored by the Snyder-Robinson Syndrome (SRS) Foundation for his pioneering research and commitment to studying this rare disease.

"Dr. Schwartz has been with the entire SRS community every step of the way," said Michael Raymond, father of a son with SRS and Executive Director of the SRS Foundation. "He's helped unlock many of the mysteries surrounding this confounding disease, and helped to coalesce a larger SRS research community. We're confident that his work will help lead to a safe and effective treatment for SRS. He has been incredibly kind, thoughtful, and compassionate in his many interactions with all of our SRS families who struggle daily with this disease."

Schwartz is a true advocate for patients, and has been often quoted as saying, "We study rare diseases that most have never heard of, but if you have a child with a rare disease, it's not rare to you, it's 100%."


Schwartz addresses guests at the opening of the JC Self Research Institute in 1

GGC's Boo Ramage shared, "No matter how rare the condition, Charles could not – and would not – shake his desire to help even that one family whose need was brought before him"

"Charles and I have worked together for almost twenty years now," said friend and collaborator, Giovanni Neri, MD, a GGC Senior Scholar and emeritus professor at Catholic University in Rome. "I have had the privilege to be witness to the monumental contributions made by Charles to the knowledge of X-linked disorders. I am highly impressed not only by the scientific quality of his work, but also by the empathy he established with patients who were the object of his research, even those he never met in person."

"Dr. Schwartz has been and always will be for me an extraordinary mentor and model," said Luigi Boccuto, MD, GGC Assistant Research Scientist. "He represents the embodiment of the scientific method: always pursuing rigor and precision, always providing constructive criticism, always giving brilliant inputs. I have had the most enjoyable and stimulating conversations with him, on both scientific and non-scientific topics, and I feel honored to have had the opportunity to work with him."

XLID EXPLAINED

Dr. Charles Schwartz's career has been dedicated to the study of intellectual disability (ID). ID affects about one percent of the population, and impacts both intellectual functioning as well as adapting to daily life. Males are more likely than females to be diagnosed with ID, mainly because there are many genes (over 140) located on the X chromosome that when altered cause X-Linked ID (XLID) (Figure 1). Dr. Schwartz and his team at GGC have been involved in identifying 30% of those genes, making GGC the international center of expertise for families and clinicians.

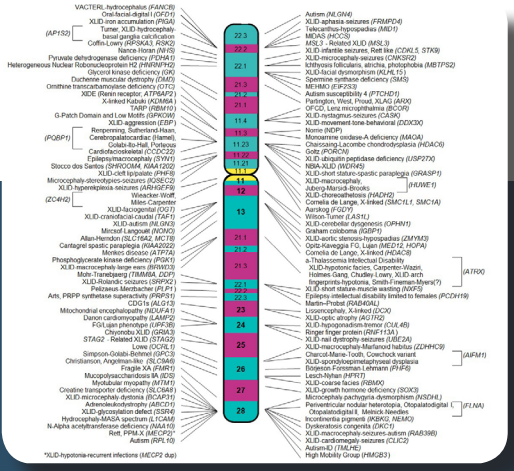


Figure 1: The X chromosome and locations of many of the XLID genes

Some of these genes cause syndromic XLID, in which patients also have other physical features, health concerns, or developmental issues. Other XLID genes cause non-syndromic ID meaning that ID is the only identifiable feature. Males have a single X chromosome, so if a gene on their X is altered, they will have the condition. Females have two X chromosomes, so even if one gene is altered, they have another copy. These conditions are typically carried by unaffected females who have a 50% chance of passing the condition onto each of their sons (Figure 2).

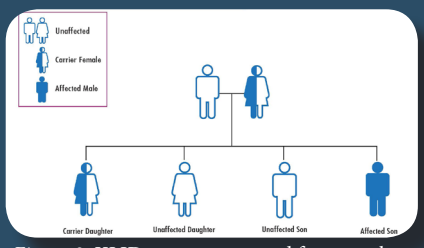


Figure 2: XLID genes are passed from mother to son - GGC Counseling Aids, 6th ed.

XLID disorders can be challenging to diagnose because, while they do often have a distinctive pattern within the family, there are so many possible genes that could be responsible. Until the gene is identified and its function understood, there is little hope of a treatment or cure. With Dr. Schwartz's accomplishments in identifying and understanding these disorders, hopes are high and work is moving ever closer to treatments that will improve the quality of life for these individuals and their families.

A SIGN OF THINGS TO COME

A new sign and logo adorn the entrance to the Greenwood Genetic Center Partnership Campus. GGC is working closely with the Greenwood Partnership Alliance (GPA), the county's economic development organization, to develop and market the campus. The vision for the project includes the recruitment of life science industry, academic partners and technologies related to human and medical genetics, biotechnology, and life science enterprises. The 190 acre Partnership Campus, owned by the GGC Foundation will also promote and support regional economic development.

With an area devoted to biotechnology growth as well as a proposed mixed-use development and space for GGC's current campus to expand, the campus is poised to offer both an environment of collaboration as well as quality-of-life benefits for prospective residents.

GPA has plans to promote the campus through various outlets and events including the BIO International Convention in Boston, Swiss Biotech Day in Basel Switzerland, and SCBIO events and meetings across South Carolina.

A website to promote the campus is expected to be completed in July.

GOLFING FOR GENES

In an annual charity golf tournament at Thornblade Club in Greenville in April, Fred Bentfeld's friends and fellow golfers surprised him with their decision that the $5,000 in proceeds from the tournament would go to the Greenwood Genetic Center Foundation in honor of Fred's granddaughter, Ella Marie Rhyne, of Greenwood (pictured below left).

Ella Marie, 18 months, was diagnosed with Kleefstra syndrome at GGC. This gift in her honor will support genetic services for other families and help advance GGC's research efforts to identify treatments for genetic disorders.

When Fred (pictured center, white shirt) shared the news with Ella's parents, Geoff and Kelly Rhyne, he said "Like me they were touched deeply. I was surprised and felt really blessed and honored to have all of these friends and know we always have their support."

"The Greenwood Genetic Center was instrumental in helping us find answers to questions we had about our beautiful Ella Marie, and they have been unbelievably supportive and informative," shared Kelly. "We are both humbled and overcome with an immense sense of gratitude."

RACE THE HELIX CONTINUES TO GROW

Race the Helix®, the signature event series of the GGC Foundation, continues to expand with larger participation each year. In March, the 5th annual Race the Helix - Upstate was held at Lake Conestee Nature Park in Greenville with over 300 participants. GGC friends, families, and interested community supporters enjoyed the beautiful morning run, breakfast, and a very successful raffle. Team Baker (left), family and friends of a young boy with metachromatic leukodystrophy showed up in full force to support the event!

The 3rd annual Race the Helix -Lowcountry was held at Palmetto Islands County Park in Mt. Pleasant on May 18th. Race hosts, Makayla Gunn, Maddie Henry, and their families (right) shared their GGC stories and welcomed supporters.

Save the Date -
Race the Helix - Greenwood - October 5, 2019

Continuing our JOURNEY OF DISCOVERY

"We are not embarking on a journey of discovery tonight," said Steve Skinner, GGC Director at the launch event for the GGC Foundation's Journey of Discovery campaign. "We have been on a journey of discovery for 45 years, but we're not on it alone. We've been on this journey with many patients and families, and we've also been on the journey with the support of many of you."

Technology advancement is the focus of the GGC Foundation's current Journey of Discovery campaign. The Center has invested $5 million in advancing three technology initiatives that faculty have deemed critical to improving patient care and remaining at the forefront of genomic medicine - genomic sequencing, model organisms (zebrafish), and confocal microscopy (see right).

The GGC Foundation Board of Trustees have committed to a fundraising campaign of $1.35 million to assist with these endeavors. The campaign runs through January 2020.

The Journey of Discovery campaign was publicly launched in April at a VIP event where faculty hosted current donors and introduced GGC's work to new friends. Attendees learned how the three new technologies that have been implemented are already being used to make diagnoses and study potential treatments.

With a focus on the collaborations between GGC's Diagnostic Laboratories and the Division of Research, Skinner; Mike Friez, PhD, Director of Diagnostic Laboratories; Heather Flanagan-Steet, PhD, Director of Functional Studies; and Richard Steet, PhD, Director of Research, highlighted the uniqueness of GGC's collaborative environment.

"The strength of GGC is that we have this tightly integrated circle that includes the clinics, where patients are seen, and continues through our diagnostic labs, through our research institute, and then back to the patient," shared Skinner. "We are unique across the country and really, around the world, in that integration."

Attendees were also treated to a guided tour of the Hazel and Bill Allin Aquaculture Facility (pictured above) which currently houses approximately 6,000 zebrafish which are model organisms for human genetic disorders. The Allins provided a transformational gift to support the facility, one of the initiatives highlighted in the current campaign.

"Unraveling the complexities of the human genome really has the potential to shape the future of healthcare," shared Steet. He noted that these three initiatives supported by the Journey of Discovery "will help place the Center at the leading edge of clinical care, diagnostic services, research, and one day, personalized medicine."

To support GGC's Journey of Discovery, contact the GGC Foundation office at (864) 388-1801 or give online.

Donate Today!

www.GGC.org/foundation

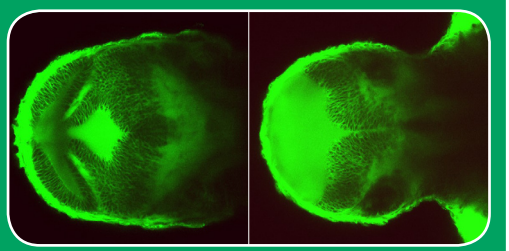
WHAT IS CONFOCAL MICROSCOPY?

GGC'S JOURNEY OF DISCOVERY SUPPORTS THIS HIGH TECH TOOL

GGC has many microscopes. We use them to analyze chromosomes, work with cell cultures, and prepare zebrafish for study. But when it comes to producing high quality imaging that is necessary to observe the finest details of development, the traditional light or fluorescent microscope is not sufficient.

Late in 2018, GGC acquired a confocal microscope that instead of using a cone of light, uses lasers to generate images. Confocal microscopy has revolutionized the ability of research scientists to follow events that occur in the cells and tissues of model organisms. The advanced technology also allows for very thin planes of a sample to be analyzed alone or combined into a 3D model.

This type of imaging allows us to see where tissue development goes awry in animal disease models without having to dissect the whole animal. Using confocal microscopy, we can: 1) look at individual cells within a disease tissue – asking how they differ from normal tissues, 2) watch a variety of developmental processes, pinpointing when and how a process diverges from normal development, and 3) use molecular tools (e.g. antibodies, probes, fluorescent proteins) to identify individual molecules and pathways disrupted during the disease process.



Above left: confocal image of the brain of a normal two-day-old zebrafish. Right: image of a two-day-old zebrafish brain that has the same genetic mutation as a GGC patient. Note the missing structures and disorganization of the affected brain. This image proved the significance of this DNA change in this child who suffers from a severe seizure disorder.

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

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