

ACCOMPLISHMENTS

- GGC's DNA Diagnostic Laboratory was one of the first labs in the world to offer clinical testing for *MECP2*, the gene associated with Rett syndrome.
- Enrollment of hundreds of patients in the natural history study through travel clinics.
- Involvement in study design, subject recruitment, protocol implementation, and data analysis on the successful trofinetide trial sponsored by Neuren Pharmaceuticals.
- Development of 'RettBe,' a neurobehavioral outcome measure.
- Publication of more than 20 peer-reviewed articles about Rett syndrome and related disorders since 2009 on topics such as diagnosis, seizures, breathing, quality of life, and survival.



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.

You can support the work of these Rett syndrome programs by contacting the GGC Foundation at www.GGC.org or 864.388.1801



Where Compassion Inspires Progress

Greenwood office
106 Gregor Mendel Circle
Greenwood, South Carolina 29646
888.442.4363

Greenville office
14 Edgewood Drive
Greenville, South Carolina 29605
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www.GGC.org/Rett

Rett Syndrome and Related Disorders

A Vision for Their Future

Programs of the Greenwood Genetic Center



WHAT IS RETT SYNDROME?

Rett syndrome is a rare, non-inherited, genetic, postnatal, neurological disorder that occurs almost exclusively in girls and leads to severe impairments, affecting nearly every aspect of the child's life: their ability to speak, walk, eat, and even breathe easily. The hallmark of Rett syndrome is near constant repetitive hand movements while awake.

-Rettsyndrome.org



"They are like family."

-Adel Forsythe, whose daughter Katy is pictured here with Dr. Steve Skinner.

The Greenwood Genetic Center was named Rett Syndrome Center of Excellence by Rettsyndrome.org in 2016!

OUR TEAM

CARRIE BUCHANAN, MD is a developmental-behavioral pediatrician at the Greenwood Genetic Center. She was awarded a Clinical Training Fellowship from Rettsyndrome.org to study behavioral disorders in Rett syndrome. Dr. Buchanan provides behavioral guidance for individuals with Rett syndrome and also participates in research and clinical trials at GGC.

STEVE SKINNER, MD is a clinical geneticist and Director of GGC. For over a decade he has led the Center's Rett syndrome natural history study team. Dr. Skinner has contributed broadly to the understanding of Rett syndrome with publications on clinical manifestations, quality of life issues, and genetic factors. Dr. Skinner is also involved in clinical trials and family support.

ALAN PERCY, MD is a Senior Genetics Scholar at GGC. He is a pediatric neurologist at Children's of Alabama and serves as Professor and Director of the Rett syndrome clinic at the University of Alabama Birmingham. He is the principal investigator for the NIH-funded Rett syndrome natural history study and is a leading clinician and researcher of Rett syndrome and Rett-related disorders.

Joining the commitment to improve the quality of life and lessen the impact of Rett syndrome and related disorders on patients and their families are:

FRAN ANNESE, LMSW - clinical research coordinator
JENNIFER STALLWORTH, MS, CGC - clinical genetic counselor
AUBIN TIERNEY - research coordinator
HANNAH WARREN, MS, CGC - clinical genetic counselor

CLINICAL CARE

The Greenwood Genetic Center is proud of our reputation for providing state-of-the-art, compassionate patient care for all of the families we serve. With a long history of expertise and interest in X-linked genetic disorders, GGC offers state-of-the-art diagnostic testing for Rett syndrome and related disorders.

More recently, the Center has established a pediatric multidisciplinary clinic based in Greenville, SC for individuals with Rett syndrome. The clinic joins specialists in genetics, neurology, developmental pediatrics, orthopedics, and physical and occupational therapy from the Greenwood Genetic Center, Greenville Health System, and Shriners' Hospital for Children. The clinic is also a place for families to become involved in clinical trials and other research studies.

RESEARCH

NIH-FUNDED RETT SYNDROME NATURAL HISTORY STUDY

- Understand the features of Rett syndrome including the course of the disorder.
- Develop "best practices" for clinical care and a foundation on which to base intervention trials.

INTERVENTION STUDIES

- Design and implementation of investigator-initiated and industry-sponsored studies with an emphasis on drug trials.
- Develop novel outcome measures using neurobehavioral assessments, biomarkers, and quality of life measures.

ADVOCACY

- Develop research agendas, white papers, and health economics documents.
- Provide leadership on key panels and committees including Rettsyndrome.org boards.