

## 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 recent successful trofinetide trial sponsored by Neuren Pharmaceuticals.
- Initial development of 'RettBe', a neurobehavioral outcome measure.
- Publication of 17 scientific articles about Rett syndrome and related disorders in the last two years on important topics including diagnosis, survival, and quality of life.



## 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](http://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  
Center for Translational Research  
14 Edgewood Drive  
Greenville, South Carolina 29605  
866.478.4363

[www.GGC.org/Rett](http://www.GGC.org/Rett)

## Rett Syndrome and Related Disorders

A Vision for Their Future

Programs of the  
Center for Translational Research (CTR)  
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



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

## OUR TEAM

WALTER KAUFMANN, MD is the Ravenel Boykin Curry Chair in Genetic Therapeutics and Director of the Center for Translational Research. From his early work in discovering that Rett syndrome is a disorder of synaptogenesis to his leadership of clinical trials, he has published extensively on Rett syndrome and has been funded through numerous major grants. In 2016, Dr. Kaufmann received the *Circle of Angels Research Award* from Rettsyndrome.org.

STEVE SKINNER, MD is the Director of the Greenwood Genetic Center and has a long-standing interest in Rett syndrome. For over a decade he has led the Center's NIH-funded 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. He received the *Circle of Angels Research Award* in 2013. Dr. Skinner is also involved in family support.

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  
LAUREN BAGGETT, MS, CGC - clinical genetic counselor  
CARRIE BUCHANAN, MD - developmental-behavioral pediatrician  
DAVID EVERMAN, MD - clinical geneticist  
JENNIFER STALLWORTH, MS, CGC - program manager  
HANNAH WARREN, MS, CGC - clinical genetic counselor



"They are  
like family."

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

## 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.
- Devise analytical strategies.
- Develop novel outcome measures using neurobehavioral assessments, biomarkers, wearable technology, and quality of life measures.

### COLLABORATIONS

- Clemson University - Big data analysis/machine learning and wearable device development to measure repetitive hand movements.
- Massachusetts Institute of Technology - Biomarker projects including blood-based markers and wearable technology.

### ADVOCACY

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