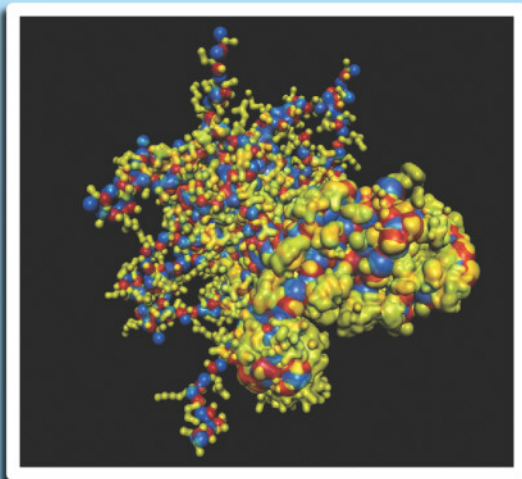


The UAB Rett Clinic, in close cooperation with the prenatal diagnostic center at Baylor College of Medicine, can guide parents on these issues.

Does RS Happen in Boys?

Most classic RS occurs only in girls, but very rarely boys are affected. New research results indicate that mutations in the *MECP2* gene can occur in a small percentage of boys and girls with other conditions. Those include non-specific mental retardation with seizures, autism, and severe neonatal encephalopathy (brain dysfunction in infants). Research is still in progress to understand the contribution of *MECP2* to these conditions better and how they relate to classic RS.



Researchers are hard at work investigating the underlying causes of RS at the molecular level.

Contact Us

Please contact us by phone or e-mail for appointment information or if you have questions about Rett Syndrome.

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www.circ.uab.edu

The UAB Team



(Shown left to right) **Alan K. Percy, M.D.**, the Medical Director for the UAB Rett Syndrome Clinic and Civitan-Sparks Clinics at UAB. **Suzanne Geerts, M.S., R.D., L.D.**, Dietician for the UAB Rett Syndrome Clinic and for Civitan-Sparks Clinics. **Jane Lane, R.N., B.S.N.**, Research Nurse Manager for the UAB Rett Syndrome Clinic. **Jerry Childers**, Program Coordinator for the UAB Rett Syndrome Clinic.

How Can You Help?

Your gift to the UAB Rett Clinic will allow us to maintain our commitment to foster discovery through basic and clinical research, to provide quality care and support for our patients, and to supply physicians with the tools they need to further their work.

You may make a contribution through an outright gift, a planned gift, or an endowed gift. To make a charitable gift or to obtain more information about giving to the UAB Rett Clinic call or email us today:

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Rett Syndrome Clinic

Civitan International Research Center



The Rett Syndrome Clinic at the University of Alabama at Birmingham (UAB) is one of the few centers in the United States that specialize in the diagnosis and care of girls and women with Rett Syndrome. The activities of the Rett Center include clinical and basic research, as well as education and promotion of Rett Syndrome awareness.

What Is Rett Syndrome?

Rett Syndrome (RS) is a disorder that occurs almost exclusively in females and was first described by Andreas Rett in 1966. However, RS did not receive international recognition until 1983. Since then, extensive research has expanded our knowledge about the clinical characteristics of RS and led to improved symptomatic treatments and quality of life for these individuals.

Typically, the girl with classic RS is born at term after an uneventful pregnancy and normal delivery. Developmentally, these girls appear normal for at least the first six months although retrospectively, some are characterized as somewhat placid or slightly floppy. The first sign is often slowing of head growth, beginning as early as 2-3 months. At 1 to 2 years, a rapid or more insidious regression occurs with loss of acquired fine motor, intellectual, and communicative abilities. They develop constant stereotypical hand movements (hand-washing or hand-wringing). Each



girl develops her own distinct, unique hand pattern. Spoken communication skills are also lost.

Following this regression phase, a period of some recovery of nonverbal communication with improved eye contact and nonverbal interactions with the environment is noted. However, a slow, insidious deterioration in motor function may occur over the next several decades. Some girls develop epilepsy, and, for many, nutritional issues and weight loss become a problem. Most girls develop a pattern of disorganized breathing characterized by periods of apnea alternating with periods of hyperventilation. This breathing pattern is present only during wakefulness and resolves during sleep. Most girls have cold feet and peripheral vasomotor disturbances suggestive of autonomic nervous system dysfunction. Other disabilities might include scoliosis (curvature of the back).

What Causes RS?

The cause of RS remained elusive until the research team of Dr. Huda Zoghbi, an investigator associated with the Baylor Rett Center, discovered that mutations in the *MECP2* gene on the X-chromosome cause RS. This discovery has opened new research avenues for the understanding of Rett Syndrome. It has led to the development of new diagnostic tests and provides a target to search for possible treatments for RS.

How Is RS Diagnosed?

It is possible to test the DNA obtained from a blood sample for mutations in the *MECP2* gene. Such mutations are found in about 95% of girls with RS. This does not mean that the other 5% do not have Rett Syndrome; we simply cannot confirm the diagnosis on a blood sample in all these girls. Further research will hopefully help us understand why this is



the case. *MECP2* testing is now routinely performed in the DNA diagnostic laboratory at Baylor College of Medicine and the Greenwood Genetic Center.

Is There a Cure for RS?

Currently no cure for RS exists. However, many treatments can be offered to girls that will improve their quality of life and that of their families. The girls should be managed through therapeutic programs that include physical therapy, occupational therapy, speech therapy with use of alternative means of communication, music therapy, hydrotherapy, and hippotherapy (horse back riding). Epilepsy can be managed with anti-epileptic drugs. Growth can be enhanced by nutritional interventions. Surgical intervention for correction of scoliosis may be necessary.

Can RS Happen Again?

In the majority of cases, RS does not recur in families. However, several families are known with more than one girl with RS. In these instances if a specific *MECP2* mutation was identified in the first girl, the second girl will have the same mutation. This means that for many parents of a girl with RS who are contemplating having another child, prenatal diagnosis in a future pregnancy can be offered as an option.

Continued ➡