Gillette Children's Specialty Healthcare

Conditions

Rett Syndrome

What Is Rett Syndrome?

Rett syndrome (also called Rett disorder) is a rare neurodevelopmental disorder that affects brain growth and development. It starts in childhood, almost exclusively in girls. Its effects on brain development can affect areas such as muscle growth, walking and communication.

What Causes Rett Syndrome?

Most children diagnosed with Rett syndrome have a gene (MECP2) that changes (or mutates) and interrupts typical brain development. This mutation causes the brain to either make too little of an essential protein, or to make damaged protein the body can't use. As a result, the brain cannot develop typically. Most cases of Rett syndrome aren't inherited.

The condition, which can produce symptoms similar to those of autism, is linked to mutations in a gene on the X chromosome. For this reason, approximately 95 percent of children diagnosed with Rett syndrome are girls. Rett syndrome in boys is extremely rare.

Rett Syndrome Symptoms and Effects

Most infants with Rett syndrome are girls who appear to develop typically until age 2 or 3. Rett syndrome symptoms appear to progress in stages. These phases vary depending on how serious the disease is, and how far along.

Early-Onset Stage

Infants with Rett syndrome usually have no symptoms during the first 6 to 12 months of life. Then, during the early-onset stage (usually between 6 to 18 months), development slows or stalls. Children stop gaining developmental skills, such as walking or talking. They might also lose skills they had achieved.

Common Rett syndrome symptoms at this stage include:

- Challenging behaviors.
- Crying or irritability.
- Difficulty crawling or walking.
- Loss of, or decline in, speech skills.
- Loss of eye contact or visual interaction.
- Loss of muscle tone (muscle weakness, floppiness).
- Lack of ability to use hands effectively.
- Repetitive hand movements (wringing, washing, tapping).
- Slowed brain and head growth.
- **Seizures**.

Experts believe the behavioral challenges linked to Rett syndrome might result from communication difficulties or an altered sensory system.

Because of its symptoms, Rett syndrome often gets mistaken for autism, **cerebral palsy** or a developmental delay. And because signs of slowed head and brain growth can be subtle, Rett syndrome might go unnoticed until other delays become obvious.

Later Stages of Rett Syndrome

As children enter their early teenage years, the effects of Rett syndrome usually slow down. Their behavior might improve, and they might cry less or be less irritable. Alertness, attention span and nonverbal communication might also improve.

However, seizures and movement problems are common. Due to immobility and muscle weakness, joint deformities and **scoliosis** might also arise. Many children remain at this stage for the rest of their lives.

Rett Syndrome Diagnosis and Treatment

Kids who have symptoms of Rett syndrome should see a **neurologist** or neurodevelopmental pediatrician for testing to determine a diagnosis.

Although there is not yet a cure for Rett syndrome, treatment can minimize its effects. As part of a comprehensive treatment plan, specialists can recommend ways to:

- Slow the loss of abilities.
- Improve or preserve movement.
- Encourage communication and socialization.

Rett syndrome treatment options might include:

- **Orthotics, Prosthetics and Seating** for custom **seating and mobility equipment** to improve or maintain independence.
- **Braces (also known as orthoses)** to help manage bone and joint deformities.
- Medicines to control seizures, reduce breathing issues, or maintain a steady heartbeat.
- **Occupational therapy** to improve or maintain functional hand use and ease repetitive hand movements.
- **Physical therapy** to improve or maintain mobility and balance.
- **Speech therapy** to help with nonverbal communication and social skills.
- Surgery for severe scoliosis or other joint deformities.

Integrated Care

If your child lives with the effects of Rett syndrome, they need the support and care of a team of experts who understand this complex condition. To give your child their best chance at leading a typical, active life, teams at Gillette Children's Specialty Healthcare organize treatment programs around the complex conditions that accompany Rett syndrome. At every stage, you and your child will work with internationally recognized experts in a family-centered environment.

When your family chooses Gillette, you have access to:

- One of the region's largest teams of complex pediatrics, pediatric rehabilitation medicine specialists, **pediatric orthopedics**, **neurologists** and **neurosurgeons**.
- Our close collaboration with groups such as the **International Rett Syndrome Foundation** and the **Midwest Rett Syndrome Research Association**.
- One of just 14 U.S. hospitals designated a Clinical Research Center of Excellence by the **Rett Syndrome Foundation**.

Additionally, girls and women who have Rett syndrome need typical primary care services. Health care providers at Gillette work with primary care doctors and gynecologists in the community to meet the overall health care needs of children and teens who have Rett syndrome.

As part of their treatment, your child might also receive care from our specialists in the following services:

- **Orthotics, Prosthetics and Seating**.
- **Cardiology.**
- **Child life**.
- **Endocrinology**.
- **Gait and motion analysis.**

- **Gastroenterology**.
- **Adult Services**.
- **Medical genetics and genetic counseling**.
- **Neurology**.
- **Neuropalliative care.**
- **Orthopedics and spine.**
- **Pediatrics and general medicine**.
- **Psychology**.
- **Pulmonology**.
- **Rehabilitation medicine**.
- **Social work**.

Drawing on all these areas of expertise, we work together to test, diagnose and develop treatment plans for each child. Our team approach lets us meet your child's needs for specialized health care, often during a single visit at one location. Our goal is to provide your family with the information you need to make informed health care decisions.

Locations

At Gillette, children under age 16 who are diagnosed with Rett syndrome usually receive treatment at our St. Paul Phalen Clinic.

Adults and teens age 16 and older receive care at our **Phalen Clinic**. We help your family determine which location is most appropriate.

Phalen Clinic

Make An Appointment **651-290-8707** Refer a Patient **651-325-2200**