

Rett Syndrome

Information for Families

If your child shows signs of or has been diagnosed with Rett syndrome, you probably have a number of questions and concerns. Our specialists who provide Rett Syndrome services understand — and we're here to help. We hope this information answers many of your questions about your child's diagnosis.

Rett Syndrome

What is Rett syndrome?

Rett syndrome is a complex neurological disorder that affects the growth and development of the brain. The syndrome produces autism-like symptoms. About 95 percent of children diagnosed with the condition are girls. An estimated one in 10,000 – 22,000 females has the condition, as do a few boys.

What causes Rett syndrome?

Between 90 and 95 percent of children diagnosed with Rett syndrome have a gene mutation (MECP2) that affects normal brain development. The mutation causes the gene to either make too little of an essential protein or to make damaged protein that the body can't use. As a result, the brain can't develop normally. The genetic mutation almost always appears spontaneously; fewer than 1 percent of recorded cases are inherited.

What are the early signs of Rett syndrome?

Rett syndrome is often mistaken for autism, cerebral palsy or a developmental delay. Early symptoms might include:

- Loss of muscle tone, which causes floppiness
- Difficulty crawling or walking
- Lack of eye contact
- Loss of purposeful use of hands
- Loss of speech
- Making repetitive hand movements
- Slowed brain and head growth
- Seizures
- Developmental delays

Some children show signs in infancy. Other children appear to develop normally until age 3 or 4.

How do doctors diagnose Rett syndrome?

If symptoms arise, doctors conduct ongoing physical and neurological (brain) evaluations. In most cases, a child's symptoms and a simple blood test to detect the mutated gene can confirm the diagnosis. A few children diagnosed with Rett syndrome don't have the mutated MECP2 gene. Such children are diagnosed based upon a clinical examination, when other possible diagnoses have been ruled out.

Do the symptoms of Rett syndrome get worse?

Rett syndrome is generally thought to progress in stages. Symptoms vary, depending on the stage and severity of the disease.

During the early stages (usually infancy to age 3 or 4), children stop gaining typical developmental skills or lose skills they'd achieved. Head growth might slow gradually, a problem that might go unnoticed until developmental delays become obvious. Children might make less eye contact and begin to lose interest in toys. They often cry and are irritable. Children can't move easily and might become unable to crawl or walk. In general, children struggle to move with purpose. Repetitive hand movements — such as wringing, washing, clapping or tapping — become more obvious.

As children become teenagers, some problems seem to lessen. Movement difficulties continue, but children often aren't as irritable. Alertness, attention span and nonverbal communication skills might improve. Other problems might worsen. Seizures are common. Secondary conditions — such as gastrointestinal, heart and breathing problems — can occur. Joint contractures or scoliosis (curvature of the spine) might arise. Severe scoliosis can prevent people from sitting upright and might affect breathing. People often stay at this stage for the remainder of their lives. In some cases, symptoms worsen again as teens become adults.

How is Rett syndrome treated?

To date, there is no cure for Rett syndrome because there is no way of supplementing the protein involved. Treatments can minimize the effects of some symptoms. For example, treatment can help slow the loss of abilities, improve or preserve movement, and encourage communication and social contact. Current research, aimed at helping us better understand Rett syndrome, could lead to drug therapies to treat the disorder itself.

At present, therapy and daily care can reduce the severity of secondary complications. Options such as medication or surgery are sometimes effective. Surgery can correct severe scoliosis. Medications help control seizures, reduce breathing problems, and maintain a steady heartbeat.

What can we expect from Gillette?

At Gillette, children, teens and adults who have Rett syndrome benefit from a team approach. Your involvement is vitally important to your child's well-being. Our team includes physicians and other specialists.

Our Core Team



Raymond Tervo, M.D., a neurodevelopmental pediatrician and medical director of Pediatrics at Gillette, is an expert in diagnostic and testing procedures. He helps families to pinpoint and understand their child's conditions.



Arthur Beisang, M.D., is a pediatrician who specializes in treating children who have complex medical conditions, such as cerebral palsy and Rett syndrome. He helps patients and families identify and gain access to appropriate providers and services. He also works to manage and lessen the symptoms associated with Rett syndrome.



Robert Wagner, M.D., is a family medicine physician and medical director of adult services at Gillette Lifetime Specialty Healthcare. He works with teenagers moving to adult medical care services and helps people manage the effects of growing older with Rett syndrome.

Because of the complex nature of Rett syndrome, our clinic patients also benefit from the services of:

- Orthopaedic surgeons
- Gastroenterologists
- Pulmonologists
- Cardiologists
- Neurologists
- Physical medicine and rehabilitation physicians

Who else might help my child?

Our team identifies other providers and services as your child needs them. Nurses are always available to help you manage your child's complex care. Other members of the team might include:

- Physical therapists, who help patients improve or maintain mobility and balance
- Occupational therapists, who help patients improve or maintain use of their hands and reduce purposeless hand movements
- Speech and language pathologists, who help patients communicate nonverbally and improve their social interactions
- Social workers, who identify community and adaptive resources to help people develop daily-living skills so they can cope with everyday needs
- Orthotists and seating specialists, who make assistive devices — such as foot orthoses and custom seating systems — to help people manage bone and joint deformities

Where can I get more information?

At Gillette, we provide medical and rehabilitation-therapy services tailored to the needs of each patient. Although Rett syndrome has no cure, our team provides supportive services that help ease associated secondary conditions. If you have questions or concerns about your child's diagnosis, please call:

- Jason Kelecic, D.P.T.
Rett syndrome services
program manager
651-312-3176
- La'Tosia Erickson, R.N.
Resource nurse
651-229-3897

To refer a patient who has or might have Rett syndrome, call 651-290-8707 or 800-719-4040, ext. 3944 (toll-free).



Gillette Children's *Specialty Healthcare*

St. Paul (Main) Campus
200 University Ave. E.
St. Paul, MN 55101
651-291-2848
800-719-4040 (toll-free)

Maple Grove Clinic
9550 Upland Ln. N.
Maple Grove, MN 55369
763-496-6000
888-218-0642 (toll-free)

Burnsville Clinic
305 E. Nicollet Blvd.
Burnsville, MN 55337
952-223-3400
866-881-7386 (toll-free)

Minnetonka Clinic
6060 Clearwater Dr.
Minnetonka, MN 55343
952-936-0977
800-277-1250 (toll-free)

Duluth Clinic
Lakewalk Center
1420 London Rd.
Duluth, MN 55805
218-728-6160
800-903-7111 (toll-free)

Mobile Outreach Clinic
For locations and schedules:
651-634-1938
800-578-4266 (toll-free)
www.gillettechildrens.org

Willmar Clinic
Lakeland Health Center
502 2nd St. S.W.
Willmar, MN 56201
651-634-1938
800-578-4266 (toll-free)



Gillette Lifetime *Specialty Healthcare*

St. Paul – Phalen Clinic
435 Phalen Blvd.
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