

# Rett Syndrome

## Background

Rett syndrome (RTT) is a genetic neurodevelopmental disorder that affects brain development. It is caused by a spontaneous mutation on the MECP2 gene, located on the X chromosome, and primarily affects females. In rare cases, males may be affected and experience a more rapid onset and progression. RTT is characterized by typical early growth and development followed by a regression in motor and communication skills, usually between 6-18 months of age.

Slowed growth and development, loss of movement and coordination, loss of communication abilities, and repetitive hand movements are characteristic of RTT. Children often develop stereotypies, which are repetitive hand movements such as wringing, squeezing, clapping, or tapping. Other signs and symptoms could include:

- Decreased head and brain growth (microcephaly)
- Muscle weakness
- Decreased verbal skills, loss of verbal ability
- Cognitive impairment
- Unusual eye movements (i.e., intense staring, blinking)
- Breathing problems (i.e., apnea, hyperventilation)
- Seizures
- Irregular heartbeat
- Scoliosis
- Decreased response to pain
- Chewing and swallowing difficulties
- Impaired stomach emptying
- Teeth grinding

There is currently no cure for RTT. Treatment focuses on managing symptoms and improving quality of life through a multidisciplinary approach. Care teams may include specialists from neurology, cardiology, orthopedics, gastroenterology, pulmonology, rehabilitative therapy, and behavioral health. Key goals of treatment include maintaining range of motion, preventing further loss of function, and supporting communication abilities. DAYBUE® (Trofinetide) is the first and only FDA-approved treatment for individuals with RTT over 2 years of age.



## Top Takeaways for School

Rett syndrome (RTT) is a complex neurodevelopmental disorder. Many children with RTT are unable to speak, walk, or use their hands purposefully as symptoms progress.

RTT is most often initially misdiagnosed as autism, cerebral palsy, or non-specific developmental delay.

Loss of purposeful hand movement increases dependence on caregivers for daily activities (i.e., inability to self-feed, interacting with toys).

Learning the student's nonverbal communication methods (i.e., gestures, vocalizing, eye gaze) is essential for building an effective communication plan with staff.

Frustration with communication is often the reason for negative behaviors. Identifying these frustrations may also reveal underlying cognitive potential.

Loss of motor function and muscle weakness may affect movement and positioning. Assess physical access and mobility in the school environment for safety (i.e., extra space, minimal obstructions).

## Considerations for the Individualized Healthcare Plan (IHP)

- Nursing diagnoses: Impaired communication, impaired thought process, risk for disturbed sensory perception and impaired physical mobility
- Nutrition interventions and equipment (consider brand/size of feeding tube, tube replacement, water flushes, fluid intake goal and supplements); note school district policy on tube replacement and consider keeping backup feeding tube kit at school if applicable
- Assessment of implanted medical device (consider location, date of surgical placement, and device-specific information)
- Use of specialized equipment, adaptive equipment, and orthotics
- Equipment troubleshooting (consider equipment/device user manual, battery, charger)
- Consider emergency action plans (EAPs) and emergency evacuation plans (EEPs) related to special health care needs, including staff education/training

## Discussion Starters for the Educational Team

1. Would the student benefit from evaluations or assessments in any of the following areas: physical therapy, occupational therapy, speech and language therapy, assistive technology, adapted physical education, functional behavior, psychology, hearing and vision?
2. Would the student benefit from additional academic support and/or modified education (e.g., copies of notes, extra time, reduced workload, simplified instructions, alternative formats for presentation of material, 504/IEP)?
3. Does the student need additional adult support to access the academic curriculum in the least restrictive environment?
4. Is the physical school environment safely accessible for the student's mobility needs (e.g., entry and exit, ramps, location of classes, access to elevator, doorways)?
5. Does the classroom environment support the student's needs and/or equipment (e.g., desk/seating options, maneuverability space, electrical outlets, flash pass for bathroom or nurse)?
6. Will staff receive education/training to implement the student-specific emergency plan?

## Resources

Kennedy Krieger Institute: Neurology and Neurogenetics Clinics  
[kennedykrieger.org/patient-care/centers-and-programs/neurology-and-neurogenetics-clinics](https://kennedykrieger.org/patient-care/centers-and-programs/neurology-and-neurogenetics-clinics)

International Rett Syndrome Foundation  
[rettsyndrome.org/](https://rettsyndrome.org/)

Rett University: IEP  
[rettuniversity.org/iep/](https://rettuniversity.org/iep/)

For more information, please scan the QR code or visit: [KennedyKrieger.org/SHNIC](https://KennedyKrieger.org/SHNIC)

