

RETT SYNDROME

Background

Rett syndrome (RS) is a genetic neurodevelopmental disorder affecting a protein that is vital for brain development. RS is caused by a random genetic mutation of the X chromosome, therefore almost exclusively occurring in female birth. RS is characterized by initial normal growth and development before a slowing or stagnation of brain development. Hypotonia, loss of function, and intellectual challenges result.

RS impairs a child's ability to speak, walk, eat, and breathe. The child can experience feeding difficulties, jerky movements of limbs, and loss of eye contact. Changes in motor skills, including loss of purposeful use of hands, are stereotypical for RS (e.g., wringing, washing, clapping hand movements). Progression of symptoms will also affect cognition, behavior, and language. Each child with RS will experience unique signs and symptoms which could include:

- Slowed growth
- Loss of purposeful hand movement, replaced by compulsive hand motions
- Loss of muscle tone
- Cognitive delay and intellectual disability
- Decreased verbal skills, loss of verbal ability
- Problems with movement, coordination, spasticity
- Difficulty chewing, swallowing, impaired stomach emptying, teeth grinding
- Skin breakdown caused by hands in mouth
- Breathing difficulties like apnea, hyperventilation
- Scoliosis
- Recurrent ear infections
- Seizures

There is no cure for RS. Treatments focus on managing symptoms, assisting with communication, and slowing the loss of abilities. Treatments may include anti-seizure medications, monitoring and treating bone health, cardiology management, feeding therapy, gastrointestinal and nutrition management, and PT/OT/SLP.



Top Takeaways for School Considerations

RS is a progressive disorder affecting neurological and motor function.

RS primarily affects expressive language, not necessarily receptive language.

The inability to communicate through speech or hand movement may underestimate cognitive ability.

Allow time for student to respond and participate in class activities. Varying forms of communication, including use of assistive technology and the eyes are useful for intentional communication.

Loss of motor function and muscle weakness may affect movement and positioning. Mobility and access to the school environment should be considered.

Considerations for the Individualized Healthcare Plan (IHP)

- Nursing diagnosis of impaired communication, impaired thought process, risk for disturbed sensory perception and impaired physical mobility
- Current diagnosed health condition including date of diagnosis, progress of disease process and other chronic health conditions
- Current medication and treatment orders (consider schedule, equipment needs and side effects)
- Student-specific triggers, avoidance, or intervention strategies
- Nutrition interventions and equipment needs (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 care plan(s) (ECP) and emergency evacuation plan(s) (EEP) as related to medical needs in the school setting, and staff education/training, as appropriate

Discussion Starters for Educational Team

1. Has the school staff been trained to implement the student-specific emergency plan?
2. 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?
3. 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)?
4. Does the student need additional adult support to access the academic curriculum in the least restrictive environment?
5. 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)?
6. 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)?

Resources

Kennedy Krieger Institute: Neurology and Neurogenetics Clinics
kennedykrieger.org

International Rett Syndrome Foundation
rettsyndrome.org/

National Organization for Rare Disorders NORD-Rett Syndrome
<https://rarediseases.org/rare-diseases/rett-syndrome/>

Rett Syndrome Research Trust
reverserett.org/



Scan QR code or visit
KennedyKrieger.org/HealthInformation for more information.