RETT SYNDROME

What is it?

Rett syndrome is a neurodevelopmental disorder caused by random genetic mutation of the X chromosome. The disease almost exclusively affects girls. Rett syndrome is characterized by early normal growth and development followed by a slowing or stagnation of brain development causing hypotonia, loss of function, and intellectual challenges. "Hands" are the hallmark of Rett syndrome. Symptoms can first include the loss of purposeful use of the hands, followed by repetitive hand movements like constant wringing, washing, tapping and clapping movements. The combination of purposeful hand use followed by loss of purposeful hand movement is required for a diagnosis of Rett syndrome. Before noticeable mental and physical symptoms appear, some subtle abnormalities may be identified. These include loss of muscle tone, feeding difficulties, jerky movements of limbs, and loss of eye contact. Often, these early signs are similar to autistic-like behaviors.

Stages

Early Onset	Occurs during ages 6-18 months. Noticeable delays in motor skills like sitting, crawling and loss of interest in toys.
Rapid destructive	Occurs during ages 1-4 years. Symptoms include loss of hand skills and language. Characteristic hand movements develop such as always bringing hands to mouth. Loss of steady gait. Breathing issues develop. Child may become irritable, frightened and reject being held. It is described as the child feeling "inside out, upside down and backwards" as they cannot make sense of their environment.
Plateau stage	Occurs during ages 2-10 years and can last for several years following. Loss of motor function and seizures are most prominent. May have improved behavior as they are less irritable, more alert, and interested in the environment. Most will remain in this stage throughout life.
Late motor deterioration	Characterized by scoliosis, muscle weakness, increased tone with abnormal posture. Hand movements may decrease.

What are the characteristics or symptoms?

- Loss of purposeful hand movement, replaced by compulsive hand motions
- Apraxia or the loss of motor function
- Skin breakdown caused by hands in mouth
- Slowed growth
- Seizures
- Cognitive delay and intellectual disability
- Breathing difficulties like apnea, hyperventilation
- Walking on toes
- Sleep problems
- Feeding difficulties like chewing
- Teeth grinding
- Loss of voice
- Altered depth perception
- Recurrent ear infections

What is the treatment?

Although there is no cure for Rett syndrome, it is important to identify and treat the different medical problems that can occur with this disorder. A team of medical and developmental specialists are key to the best possible outcome. Those diagnosed with Rett syndrome will often be followed by numerous specialists which might include neurologists, gastroenterologists, pulmonologists, orthopedists, and clinical geneticists. The pediatrician will often manage the multidisciplinary approach. Treatments might include: feeding therapy, anti-seizure medications, and PT/OT/SLP.

Kennedy Krieger Institute

The **Specialized Health Needs Interagency Collaboration (SHNIC)** program is a collaborative partnership between the Kennedy Krieger Institute and the Maryland State Department of Education.

Suggested school accommodations

Rett sydrome has a wide variety of symptoms and the severity of the symptoms can vary. Rett syndrome is a progressive disorder that affects motor and neurological functions and a student can regress quite rapidly and lose previously acquired skills. Supporting students with this condition in the school require educators and parents/guardian to work as a team. Some accommodations to consider for a 504/IEP could include:

- PT/OT/SLP evaluations
- Assistive communication devices
- Placing object choices on up/down plane vs. side by side
- Temperature control in all settings
- Executive function strategies
- Multiple teaching modalities and strategies due to response time, and method of communication
- Extended time
- Expect participation and allow time for responses
- Provide repetition of activities
- Staff education/training as appropriate
- Emergency Evacuation Plan (EEP)

Specific health issues for Individualized Healthcare Plan

- Diagnosis including other symptoms/disorders
- Documentation for type of seizure, characteristics, length of, warning signs, triggers, etc.
- Current medication list for both home and school
- Order for emergency medications including when to administer, dose, route, time
- Order for hidden device like a vagus nerve stimulator,
- Nutrition orders including fluid intake goals and need for any thickeners (drooling may add to lose of fluids)
- Nutrition orders including diet and route for meals and snacks (Example: Ketogenic diet, feeding tube, etc.)
- Note formula, back up formula, water flushes, replacement of feeding tube per school district policy
- Orders for braces/orthotics
- Baseline skin assessment
- Note student's high pain tolerance and how to properly assess pain including use of pain scale
- Communicate with school staff, parents, and provider any changes or concerns about the disorder
- Emergency Care Plan(s) (ECP) related to medical needs in the school setting and staff education/training as appropriate for each

Resources & Manuals

Kennedy Krieger Institute: The Neurology and Neurogenetics Clinic
https://www.kennedykrieger.org/patient-care/centers-and-programs/neurology-and-neurogenetics-clinic

International Rett Syndrome Foundation
https://www.rettsyndrome.org/

National Institute of Neurological Disorders and Stroke
https://www.ninds.nih.gov/Disorders/All-Disorders/Rett-Syndrome-Information-Page

Rett Syndrome Research Trust
https://reverserett.org/