

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 that causes hypotonia, loss of function and intellectual challenges. "Hands" are the hallmark of Rett syndrome and symptoms can first include the loss of purposeful use of the hands. Repetitive hand movement then follows with 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.



Symptoms

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

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 stage: During ages 1-4 years. Loss of hand skills and language. Characteristic hand movements develop like 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.

Senses

Sight: Altered depth perception and orientation to space. This may make going down steps impossible.

Sound: A small percentage of children may have sound perceptions affected by recurrent ear infections. They can become irritated or frightened with noisy environment.

Touch: Do not like face being touched but will often rub their own face and eyes. Prefer room temperature food and texture is important. Will swallow solids more easily than liquids.

Taste: Have likes and dislikes just like any other child.

Smell: Seems to be normal in children with RS.

Suggested school accommodations

- Alternative or assistance communication devices
- Develop choice making
- Placing object choices on up/down plane vs. side by side
- Using flash cards
- Communication boards
- Calendar boards
- Music therapy
- “Don’t say it, sing it”
- Offer one task at a time
- Expect participation and allow time for responses
- Provide repetition of activities

Techniques for classroom

For calming

- Slow, rhythmic movement
- Simple melodies, low tone
- Weighted vest or blanket
- Warm temperatures
- Unchanged visual stimuli
- Dim light
- Firm stroking or compression

For arousal

- Jerky movement, change in direction
- Light touch
- Cold temperature
- Bright lights
- Red/Yellow color shades
- Black/white patterns
- Touching face

SHNIC school nurses information:

Specific health issues for individual health care plans

- 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, feeding tube
- Seizure action plan, when to call 911
- Return to class protocol following a seizure
- 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

Resources & Manuals

International Rett Syndrome Foundation

<https://www.rettsyndrome.org/>

National Institute of Neurological Disorders and Stroke

http://www.ninds.nih.gov/disorders/rett/detail_rett.htm

Rett Syndrome Research Trust

<https://reverserett.org/>