Rett/ Rett variant Syndrome (MECP2- Related Disorders)
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools (www.gemssforschools.org).

Rett Syndrome is one of several conditions involving the MECP2 gene. The MECP2 gene is on the X chromosome, and therefore the manifestations of these conditions are different in boys and girls. For a discussion of MECP2 Duplication in males see: https://www.gemssforschools.org/conditions/mecp2-duplication/default.

Physical characteristics and/or symptoms

Note: not all people with Rett/Rett variant syndrome will have all of these features.

Classic Rett syndrome

Classic Rett syndrome is a progressive neurodevelopmental disorder primarily affecting girls. It is characterized by:

- Normal psychomotor development during the first 6-18 months, followed by a short period of developmental stagnation, then a rapid regression of language and motor skills, followed by long term stability.
    - Abnormal muscle tone, which may lead to muscle spasms and abnormal posture
    - Foot and hand deformities
    - Repetitive, stereotypic hand movements that replace purposeful hand use
    - Fits of screaming and inconsolable crying
    - Autistic features
    - Panic-like attacks
    - Teeth-grinding
    - Irregular breathing
    - Instability when walking or abnormal gait
    - Tremors and seizures (90%)
    - Acquired microcephaly (small head size)
    - Scoliosis/kyphosis (80%)
    - Diminished response to pain
    - Small, cold hands and feet

- Bowel dysmotility, constipation
- Unusual eye movements
- Reduced bone mass (74%)
- Growth failure and wasting that worsens with age (80-90%)

Atypical or variant Rett syndrome

- Intellectual disability with abnormal increase in muscle tone, muscle stiffness, or muscle tremors
- Age when symptoms first appear varies
- Some females with MECP2 mutations present with what appears to be isolated autism or mild learning disability (rare)

Affected males

- In males with Classical Rett gene changes, severe neonatal brain disease occurs, usually resulting in death before age 2
- **NOTE: MECP2 duplications cause a separate condition that affects males**

Recommended Routine Surveillance

- Assessment of feeding/eating, digestive problems (including constipation and gastroesophageal reflux)
- Surveillance for scoliosis
- Monitor for sleep and/or breathing problems
- Video/EEG monitoring to monitor for seizures
- Screening for prolonged QTc by ECG and Holter monitoring

Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.

- Emergencies should be handled as with any child.
- If seizures are present, the following seizure action plan may be useful:

  https://www.aap.org/en-us/Documents/Seizure_Action_Plan_for%20School.pdf

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child's individual needs is recommended.

- Cardiologist
  - Screening for prolonged QTc by ECG and Holter monitoring
- Developmental evaluation
  - Speech therapy with emphasis on nonverbal methods of communication
  - Physical therapy
  - Occupational therapy
- Gastroenterologist
  - Gastroesophageal reflex
  - Constipation
- Geneticist / Genetic Counselor
  - Diagnosis
  - Coordination of care
  - Genetic risk for family
  - Clinical trials
- Neurology
  - Follow with appropriate MRI and EEG
  - Monitor seizures
- Orthopedists
  - Gait ataxia
  - Scoliosis
  - Muscular hypotonia

Sample Forms

Sample paragraph to be used for Letters of Medical Necessity or Letters to the school
My patient_____ has been diagnosed with Rett syndrome. Rett is a neurodevelopmental disorder primarily in girls. It is characterized by developmental delay and regression, repetitive and stereotypic hand movements, autistic features, ataxia, seizures, and acquired microcephaly. Medical complications with Rett syndrome include management of seizures, scoliosis, prolonged QT, gastrointestinal reflux, and constipation. Individuals may also lose their speech and ability to walk. Because of these, _____ needs the following accommodations.

Seven Important Aspects of School Life

"Rett/Rett variant Syndrome at a Glance" will help you talk with parents and schools about:

- Medical / Dietary Needs
- Education Supports
- Behavior & Sensory Supports
- Physical Activity, Trips, Events
- School Absences & Fatigue
- Emergency Planning
- Transitions

Resources

Gene Reviews
http://www.ncbi.nlm.nih.gov/books/NBK1497/

Genetic Home Reference
https://ghr.nlm.nih.gov/condition/rett-syndrome