Rett/ Rett variant Syndrome (MECP2-Related Disorders) At-a-Glance

Rett syndrome is one of a group of disorders known as the MECP2 related disorders, caused by a change, or mutation, in the MECP2 gene on the X chromosome. This group of disorders includes classic Rett syndrome, variant Rett syndrome mild learning disabilities, and rarely, autism. These conditions occur almost exclusively in females. When the same gene change occurs in a male it is usually either lethal or causes a severe neurological condition at birth.

About 1 in every 8,500 baby girls born has an MECP2 related disorder by 15 years of age.

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.
  - However, most girls will likely develop dystonia, and foot and hand deformities as they grow older.
  - During the rapid regression, individuals will have 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 to walk or abnormal gait
  - Tremors and seizures (90%)
    - Generalized tonic clonic seizures and partial complex seizures are most common
  - Acquired microcephaly
  - Scoliosis/kyphosis (80%)
  - Diminished response to pain
  - Small, cold hands and feet
  - Bowel dysmotility, constipation
  - Unusual eye movements
    - Intense staring
    - Blinking
    - Closing one eye at a time
  - Osteopenia (74%)
- Can lead to an increase in risk of fractures
- Ambulatory individuals have better bone density than non-ambulatory individuals
  - Growth failure and wasting that worsens with age (80-90%)
  - May be caused in part by poor food intake

**Atypical or variant Rett syndrome**

- Intellectual disability with spasticity or tremor
- Variable onset
- Mild learning disability (rare)
- Autism (rare)

**Affected males**

- In males, severe neonatal encephalopathy occurs usually resulting in death before age 2

**Things to Think About**

**1. Medical / Dietary Needs**

**What you need to know**

The list of possible medical problems in Rett/Rett variant or MECP2 related disorders can be quite extensive. However, each individual usually has only some of these problems. Also, the severity of any one of these medical problems varies widely. Therefore, it is important to ask the parents about the medical issues for their child.

Treatment is mainly symptomatic and multidisciplinary, and should include psychosocial support for the family.

A dietician maybe involved

- Increased fluid intake and high fiber diet can help prevent acute intestinal obstruction. Miralax, and stool softener may be used.
- Anti-reflux agents, smaller amounts, thickened feedings, and positioning can help with decreasing gastrointestinal reflux.
- Bone loss may occur so-careful attention to nutrition and calcium intake is important.
- Some children may use a ketogenic diet or L-carnitine.

**What you can do**

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• Report any change in seizure activity to the parents. Follow school protocols when seizures do occur.
• Ensure a yearly check up in the child’s Medical Home.
• Ensure up to date immunizations. Most children with Rett/Rett variant or MECP2 related disorders can receive live virus vaccinations. Record types of vaccinations the child receives.
• Support good hand washing to reduce the spread of viruses.
• Notify parents of changes in energy level.
• Be aware of any changes in behavior or mood. Notify the parents.
• Be aware of any changes in academic performance. Contact parents.
• Be an advocate for the child who uses communication supports so that the child can communicate effectively throughout the day. This may include alternative and augmentative communication systems or devices.
• Periodic cardiac evaluation to monitor for changes in heart rhythms.
• Dietary: GERD (gastroesophageal reflux) may occur. Talk with the parents about particular foods that might be triggers for the reflux and avoid those foods. If the child has more vomiting or reflux than normal, contact the parents so that the cause can be determined.

2. Education Supports

It is important to have HIGH EXPECTATIONS for learning for children who have Rett/Rett variant or MECP2 related disorders. Encourage use of the core educational curriculum and modify how it is taught in order to meet the individual needs of the child.

What you need to know

Girls who have Rett/Rett variant or MECP2 related disorders may have

• Abnormal muscle tone
  o Motor milestones that may be delayed include:
    ▪ Sitting and crawling
    ▪ Walking
    ▪ Some have ataxia gait
  o Hypotonia leads to spasticity (75%)
    ▪ Often more pronounced in legs and may lead to mild contractures over time
• Disturbances in blood flow, especially in lower limbs
• Intense eye communication or use eye pointing as part of their communication
• Music therapy, therapeutic horseback riding, swimming have been reported to be of benefit
• Cognitive challenges of varying degrees, with or without seizures
• Very delayed speech development; majority of children do not develop speech

Environment

Consider what affects the environment may have on the child and what offers most comfort.
• Lighting

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• Noise level
• Position
• What stresses her
  o Address issues such as when the child is flooded with sensory overload, is fatigued, or has anxiety

**Eye gaze devices**

• My Tobii might offer technology to assist

**What you can do**

**Motor Issues**

• Ataxia (difficulty coordinating smooth motor movement)
  o Unstable or non-walking children may benefit from physical supports in the classroom.
  o They may need extra supports/people to help them in their academic program and to be fully included.
  o Children who have Rett/Rett variant or MECP2 related disorders with more motor issues may need extra space and/or minimal obstructions to be safe.
• Physical therapy
  o Adaptive chairs or positioning support may be helpful
  o Stretching exercises can help maintain joint range of motion, prevent secondary contractures, and prolong ability to walk
• Occupational therapy
  o May help with fine motor and oral motor control
  o Sequencing may be hard due to fine motor challenges
  o Potential as oral speakers
• Movement
  o Physical, speech, and occupational therapy to enable walking, proper positioning, hand use, communication needs, etc.
  o Bracing and surgery may be needed to align legs
  o Ensure all areas are safe, free of obstacles

**Appropriate educational strategies**

Individualized, flexible, and appropriate educational strategies/supports are keys to success.

• Intellectual ability may be underestimated due to lower functional abilities.
  o Developmental testing may be difficult because of attention, activity, speech and motor issues.
  o Formalized testing has limitations. Make sure testing consists of observations in natural settings
• Attention
  o They may pay more attention when they are naturally curious

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High interest in communication is a sign that child is ready to learn sign language and other ways to communicate.

Speech and Language

- Language challenges are significant in Classical Rett syndrome.
  - A small number of children with Rett will learn to use 1 or 2 words consistently
  - May communicate by pointing, using gestures, and using communication boards and AAC devices
  - When children have difficulty communicating, they may resort to behaviors such as pulling hair, hitting, biting. Suggestions for therapy:
    - Assess if augmentative communication device is necessary and/or appropriate for therapy.
    - Frustration with communication is often the reason for negative behavior. Identifying and intervening to reduce communicative frustrations through environmental modifications is suggested.
    - May not need a behavior plan but rather an effective communication system
  - Maintain high expectations as abilities vary widely, particularly in Rett variant/MECP2-related disorders. Always assume competence when beginning speech and language therapy.

- Teach learning strategies for non-verbal expression.
  - Consider new technology, computers, and possibly sign language depending on fine motor skills. Focus on non-verbal methods of communication.
  - Use augmentative communication aids such as picture cards or communication boards early.
  - Communication should work with child’s desire to socially interact with others in natural settings.
  - Make sure children have opportunities for choice and control in their lives.
  - Incorporate typically developing peers into their therapy to promote social interaction as well as provide typical models of language.
  - Find AAC system that allows for maximal social reciprocal communication
  - Encourage finger pointing early to help with device use as they age
  - Anyone interacting with the child should have education and training on how to encourage reciprocal communication with the device
    - Modeling is important for learning to use of the AAC device and to encourage its use.
  - Continue with strategies that improve oral control to maximize their communication

3. Behavior and Sensory Supports

What you need to know

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Individual with Rett/Rett variant or MECP2 related disorders related disorder should have neuropsychological evaluations to assess abilities and offer support for behavior challenges.

**Behaviors**

- Seemingly inappropriate laughing/screaming spells
- Sudden, odd facial expression and long bouts of laughter
- Screaming that occurs for no apparent reason
- Hand licking
- Grasping of hair or clothing
- Increasing agitation and irritability as they age

**Social**

- Social opportunities allow children to express a broad range of feelings and form close bonds and real friendships with others.
- They should be part of typical family and class activities, household chores, and perform daily living skills.
- Learn their interests and preferences.
- Behavioral supports may be helpful in limiting the less desirable behaviors that are socially disruptive and/or self-injurious.

**What you can do**

**Appropriate teaching strategies**

- Make sure teaching strategies being used are appropriate for children who are already socially engaged.

**Behavioral supports**

Be proactive with behavioral supports.

- Discuss involvement of behavioral or mental health professionals, and/or medications with the parents as needed.
- Firm directions, rules, and clear expectations are helpful.

**Regulating emotions and behavior**

Many children have difficulty regulating emotions and behavior. This is especially true when handling unplanned changes.

- Talk through expected changes.
- They usually thrive with consistency and routine. They can be easily upset with disruption.
- Prepare for any change in schedule.
- Provide a safe area to share emotions.
- Teach and model use of words and/or pictures in sharing emotions.
• Teach, emphasize, and reinforce behaviors you want to see.
• Make sure they have an effective communication system.

**Social cues and coaching**

Provide social cues and coaching.
• Provide information to and discuss differences with the child’s peers.
• Help develop confidence and focus on strengths.
• Provide positive reinforcement.
• Teach how to recognize facial expressions, body language, and moods in others.
• Teach how to regulate own body – sensory strategies may be helpful.

**4. Physical Activity, Trips, & Events**

**What you need to know**

• Any change in routine may produce anxiety, fears, and/or worry. Crowds and loud noise may be hard for some children.

• If you live in New England (USA) and qualify, Northeast Passage offers Therapeutic Recreation and Adaptive Sports programming ([www.nepassage.org](http://www.nepassage.org)).

**What you can do**

• Be proactive and discuss any change in schedule or setting with the child ahead of time.
• Use social stories and pictures to help them understand the change.
• Encourage use of their communication system to help them process concerns.

**5. School Absences and Fatigue**

• Children with Rett/Rett variant or MECP2 related disorders may be absent due to illness and/or medical appointment
  o Help to make transitions in and out of school as seamless as possible
• Children with Rett/Rett variant or MECP2 related disorders may be tired and require rest opportunities or breaks in their day.
• They may have an impaired sleep pattern.
  o Melatonin may be used to help with sleep disturbances

**6. Emergency Planning**
What you need to know

Emergency plans will be individually determined, based on behaviors and medical issues. It is important to mention new signs, symptoms, or pain to the child’s parents.

7. Resources

**Genetic Home Reference**

*Your guide to understanding Genetic Conditions*

**National Center for Biotechnology Information – MECP2 Related Disorders**

http://www.ncbi.nlm.nih.gov/books/NBK1497/
*Learn more about the genetics of MECP2 Related Disorders*

**Rett Fact Sheet**

*from the National Institute of Neurological Disorders and Stroke (NINDS)*

**International Rett Syndrome**

www.rettsyndrome.org
*The core mission of the IRSF is to fund research for treatments and a cure for Rett syndrome while enhancing the overall quality of life for those living with Rett syndrome by providing information, programs, and services.*

**Rett Syndrome Research Trust**

www.rsrt.org
*A cure for Rett Syndrome – it’s our obsession*

**Girl Power 2 Cure, Inc.**

http://www.girlpower2cure.org/Home.aspx
*We are committed to making Rett Syndrome the first reversible neurological disorder by harnessing the spirit of girls to support fellow girls who are suffering. We support girls in the planning and implementation of events that raise awareness and funds for Rett Syndrome research, as well as support Rett families with resources, fundraising help and awareness tools.*

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