

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.



Meet Jocelyn on page 9

About 1 in every 8,500 baby girls born has an MECP2 related disorder by the time they turn 15.

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%)
 - Generalized tonic clonic seizures and partial complex seizures are most common
 - Acquired microcephaly (small head size)
 - 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
 - Reduced bone mass (74%)

- Increased risk of fractures can occur
 - 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 abnormal increase in muscle tone, muscle stiffness, or muscle tremors
- Age when symptoms first appear varies
- Mild learning disability (rare)
- Autism (rare)

Affected males

- In males, severe neonatal brain disease 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 may be involved.

- Increased fluid intake and a high fiber diet can help prevent acute intestinal obstruction. Miralax and stool softeners 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 (a high-fat, adequate-protein, low carbohydrate diet) or L-carnitine supplements.
- Be aware, or ask a parent, if the child has a medical alert bracelet.

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 immunization records are up to date. 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 if the child uses communication supports so that the child can communicate effectively throughout the day. This may include alternative and augmentative communication systems or devices.
- Ensure child is having periodic cardiac evaluations to monitor for changes in heart rhythms.
- Dietary: GERD (gastroesophageal reflux disease) may occur. Talk with the parents about particular foods that might be triggers 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 LEARNING EXPECTATIONS for children who have Rett/Rett variant or MECP2 related disorders. Encourage use of the core educational curriculum and modify it in order to meet the individual needs of the child.

What you need to know

Possible issues

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

- Abnormal muscle tone
 - Motor milestones that may be delayed include:
 - Sitting and crawling
 - Walking
 - Some have ataxia (difficulty coordinating small motor movement) gait
 - Low muscle tone can lead to abnormal increase in muscle tone, muscle stiffness (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
- 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
- Noise level
- Position
- What stresses her
 - 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)
 - Unstable or non-walking children may benefit from physical supports in the classroom.
 - They may need extra supports/people to help them in their academic program and to be fully included.
 - 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
 - Adaptive chairs or positioning support may be helpful.
 - Stretching exercises can help maintain joint range of motion, prevent secondary contractures, (abnormal shortening of the muscle that can result in deformity or distortion) and prolong ability to walk.
- Occupational therapy
 - May help with fine motor and oral motor control
 - Planning and coordinating physical movements may be hard due to fine motor challenges.
 - Evaluate a person's potential as an oral speaker
- Movement
 - Physical, speech, and occupational therapy to enable walking, proper positioning, hand use, communication needs, etc.
 - Be aware that bracing and surgery may be needed to align legs
 - Ensure all areas are safe, free of obstacles
 - Music therapy, therapeutic horseback riding, and swimming have been reported to be of benefit

Appropriate educational strategies

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

- Intellectual ability may be underestimated due to lower functional abilities.
 - Developmental testing may be difficult because of attention, activity, speech and motor issues.

- Formalized testing has limitations. Make sure testing consists of observations in natural settings
- Attention
 - Child may pay more attention when they are naturally curious.
 - 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 Alternative and Augmentative Communication (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 the 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 the child's 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 so as to develop and maintain the child's ability to use devices.
 - 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 the AAC device and to encourage its use.
 - Continue with strategies that improve oral control to maximize the child's communication

3. Behavior and Sensory Supports

What you need to know

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

Behaviors

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

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.

- The child usually thrives with consistency and routine. The child 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 the child has 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).

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 appointments.
 - 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.
 - 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

Medline Plus

<https://medlineplus.gov/genetics/condition/rett-syndrome/>

Consumer-friendly information about human genetics from the U.S. National Library of Medicine

GeneReviews

<http://www.ncbi.nlm.nih.gov/books/NBK1497/>

Learn more about the genetics of MECP2 Related Disorders

Rett Fact Sheet

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

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

International Rett Syndrome Foundation

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.

Classroom Accommodations for Students with Visual Issues

Classroom Accommodations for Students with Visual Issues is a resource from Boulder Valley Vision Therapy, P.C. (<https://www.bouldervt.com/>), in Boulder, Colorado. It describes a variety of variety of issues and offers strategies.

<https://www.bouldervt.com/wp-content/uploads/sites/478/2015/12/227-Course-Handout-CCIRA-2016.pdf>

Meet a young woman with Rett – Jocelyn’s Journey

Jocelyn is a woman who is living independently and has supportive roommates, family and friends helping her live an active life as an adult in her community.



Jocelyn began her public school journey in the early 1980’s in her small city in New Hampshire. Until fourth grade, she was entirely in special education classes. After her mother attended the NH Leadership Series, she asked the school about including Jocelyn in more meaningful ways in typical classes. Her request was met with enthusiasm, and, in fact, one teacher requested that Jocelyn be in her class! As with many programs in the 1980’s the social component of school was valued quite highly, while academic expectations were not as high. That has changed!

Jocelyn’s social life was extremely successful. In fact, she and her friends were the subject of a movie on friendships called “Voices of Friendship” which is a great testament to the power of friendship! (See below for more information on the Video).

Jocelyn worked hard with her team to come up with a communication system that was effective but that didn’t happen for quite a few years. She had the services of an Inclusion Facilitator while in school to help her school experience be more inclusive in all aspects of education.

Jocelyn graduated at age 18 with her peers and began her post graduate experiences. She had the services of a Home Health person who helped her attend classes at the Tech school, enjoy swimming at the pool, and volunteer in her community. She met several people who helped her

engage more fully in college classes, including educators who valued Jocelyn’s educational and life experiences and sought her as a co-teacher in their university courses. With her mother’s support, she prepared content that went along with topics on the syllabus and presented them to the class.

Learn more details of how Jocelyn taught, from Jocelyn’s mother:

I helped Joce prepare her presentations by sharing her experiences and asking her lots of questions (yes / no) to get to something she was happy with that shared her feelings and story.

The first year I wrote her story on a paper and it was read by a student. The next year I purchased a used laptop and wrote her presentation in a reader program (Read Please, as suggested by Karen Erickson), I would set up her computer to a LCD projector in the class and when she was ready to present, the computer read it aloud to the class while the projector showed the text. Jocelyn would be able to interact with the students 1 to 1 or in small groups by responding to their specific questions (yes / no or when given clear choices).

This continued for several years, thru 2 professors (Susan Shapiro and then Cheryl Jorgenson) and a total of 4 different class topics (Best Practices In Elementary School; Best Practices In Secondary School; Contemporary Issues For Persons With Developmental Disabilities; and Facilitating Relationships). The last classes were even online classes.

I supported her through help writing her presentations but only supported her in class itself for the facilitating relationships classes.

Later, as service dollars increased and she became eligible for subsidized housing, she moved into her own home which she rents using her funding streams. She has friends and paid supports who help Jocelyn have a full and engaged life. She has been to a Singles Club, goes to movies, shops and hangs out with friends. Her family lives locally and provides support as needed to fill any gaps and Jocelyn goes to her parent’s home every other weekend.

Her transition from pediatrician to adult family practitioner came at about age 21. Her parents have power of attorney over her medical and financial needs but Jocelyn is her own guardian. She makes many decisions using an eye gaze and yes/no system. She reads people very quickly and is very sensitive to fairness, justice, and wanting to make her own decisions.

Her mother’s advice for teachers is to:

- “assume competence”
- challenge themselves to think of the big picture
- make sure their student’s know that they believe in them

Her advice to families is to:

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- Don't hold back
- Let your kids make mistakes and don't be afraid of something going wrong
- Share responsibility with others
- Get your child set up independently and work out the kinks
- Appreciate that as her daughter gained independence, her life blossomed and became richer!

View Voices of Friendship on YouTube: <https://www.youtube.com/watch?v=g6Yp4MzNQMk>