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Genetic Education Materials for School Success

Rett Syndrome

(MECP2-Related Disorders)

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Acknowledgment:

Genetic Education Materials for School Success (GEMSS) was created and maintained by an interdisciplinary team of genetic counselors, physicians, families, and students working with New England Regional Genetics Network (NERGN) and the Institute on Disability at the University of NH. Thank you to these amazing partners. This work was supported by HRSA grant UH7MC30778.

Disclaimer:

This is not intended to be medical or clinical advice. Any recommendations here should be reviewed for the appropriateness for your individual child and their unique situation. Also, please note that each document should include the date that it was last reviewed or updated. Research and guidelines that have been released after that date may represent more up-to-date recommendations.

Rett Syndrome

(MECP2 - Related Disorders)

At a Glance

Meet Jocelyn on page 11

Rett syndrome is a rare genetic condition caused by a change in the MECP2 gene on the X chromosome. It is characterized by apparently normal development during infancy followed by a loss of developmental milestones. There are different forms of this disorder which vary in presentation. Children with Rett Syndrome are almost always born to unaffected parents meaning there is no family history. This condition occurs almost exclusively in females.

Common Features of Rett Syndrome

- Slow development
- Small head size
- Apraxia - difficulty coordinating motor functions, e.g., speaking, controlling eye movements
    - Loss of function in the hands replaced by distinctive hand movements
- Difficulty walking – from wheelchair dependent to unusual gait (e.g., walking on toes, or wide-based gait
- Seizures
- Cognitive problems
- Digestive problems
- Breathing difficulties while awake
    - Breath holding
    - Hyperventilation
    - Swallowing air
- Sleep problems
- Teeth-grinding
- Difficulty chewing

Created by NERGN with funding from HRSA grant UH7MC30778. Revised 5/2024

In Depth

Medical and Dietary Considerations

What you need to know

The list of *possible* medical problems in Rett Syndrome 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 medical issues for their child.

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.

Possible Conditions Seen in Rett Syndrome

- Abnormal muscle tone, which may lead to muscle spasms, contractures 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
- Microcephaly (small head size)
- Scoliosis/kyphosis (curved spine)
- 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 which increases risk of fractures
- Growth failure and wasting that worsens with age
    - May be caused in part by poor food intake

While there is no cure for Rett syndrome, in 2023, the U.S. Food and Drug Administration (FDA) approved a new drug, *Trofinetide,* to treat Rett syndrome in children aged two and older. Trofinetide works by reducing swelling in the brain, increasing the amount of a protective protein, and stopping some cells from becoming too active.

What you can do

- Encourage a yearly check-up in the child's Medical Home. Obtain any updated care plans.
- Ensure immunization records are up to date.
- Support good hand washing to reduce the spread of viruses.
- Anti-reflux agents, positioning, and smaller amounts of thickened feedings can help with decreasing gastrointestinal reflux (GERD).
- Talk with the parents about particular foods that might trigger GERD and avoid those foods. If the child has more vomiting or reflux than normal, contact the parents so that the cause can be determined.
- Increased fluid intake and a high fiber diet can help prevent acute intestinal obstruction. Laxatives and stool softeners may also be used.
- 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 of any changes in behavior, mood, or energy level. Notify the parents.
- Report any change in seizure activity to the parents. Follow school protocols when seizures do occur.

Education Supports

It is important to have high learning expectations for children who have Rett/Rett variant or MECP2 related disorders, utilizing their strengths and interests as a foundation. Encourage use of the core educational curriculum and modify it to meet the child's individual needs.

What you need to know

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

- Abnormal muscle tone
    - Motor milestones that may be delayed – sitting, crawling, walking
    - Some have ataxia (difficulty coordinating small motor movements and gait
    - Can lead to increased muscle tone and muscle stiffness
        - Often more pronounced in legs, 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

What you can do

Consider what effects the environment may have on the child and what offers the most comfort.

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

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 the child is ready to learn sign language and other ways to communicate

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

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 and provide typical language models
    - Find alternative and augmentative communication systems or devices that allow for maximal social reciprocal communication
    - Encourage finger pointing early 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

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
- When children have difficulty communicating, they may resort to behaviors such as hair pulling, hitting, and/or biting
- Any change in routine may produce anxiety, fears, and/or worry. Crowds and loud noise may be hard for some children

What you can do

Behavioral Supports

Be proactive with behavioral supports

- Discuss involvement of behavioral or mental health professionals, 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 Support

- 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, classroom jobs, 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

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 self-regulate – sensory strategies may be helpful

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

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

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 have an impaired sleep pattern. They may be tired and require rest opportunities or breaks in their day.

Emergency Planning

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.

Resources

Medline Plus

MedlinePlus is an online health information resource for patients and their families and friends. It is a service of the National Library of Medicine. This link will provide a good medical overview of the condition without being too technical. https://medlineplus.gov/genetics/condition/rett-syndrome/

International Rett Syndrome Foundation

World-wide network of parents, clinicians, and researchers. It has numerous resources for families who are at every level of the Rett journey.www.rettsyndrome.org

Girl Power 2 Cure, Inc.

This organization started by a mother of a daughter with Rett Syndrome has many helpful resources including "Rett University." http://www.girlpower2cure.org/Home.aspx.
Check out the page with information about drafting IEP. https://rettuniversity.org/iep/

Rett Syndrome Research Trust

Focused on funding and supporting research for Rett Syndrome. www.rsrt.org

Info for School Nurse and Primary Care Staff

Rett Syndrome (MECP2 related disorders) is a complex condition that will be best managed by good collaboration between the school nursing staff and primary care staff as part of the student's team. Work with the parents to get signed releases to share information at the school nurse's office and the primary care office.

Gene Reviews is a peer-reviewed point-of-care reference accessible via the National Library of Medicine. The entry for Rett Syndrome (MECP2 related disorders) provides a comprehensive summary of the condition and describes some ongoing surveillance and management issues. It is a great resource for medical providers. https://www.ncbi.nlm.nih.gov/books/NBK1497/

HIPAA Concerns

Sometimes concerns by primary care staff about violating HIPAA regulations can hamper care by limiting access to necessary medical information. Please note that if care is intended to be delivered by school staff, clarifying medical orders or medication instructions represents continuation of medical care and is not a violation of HIPAA. To avoid any issues like this, remind the parents to sign a release of information form at the primary care office. The school nurse should keep a copy of that release in the student's file, if possible, to facilitate sharing of information in the event that there is difficulty obtaining needed information.

Example letter for physician to provide to school

"My patient______________________ has been diagnosed with Rett Syndrome. This is a rare genetic condition that is characterized by developmental delay, neuromuscular issues, speech and learning difficulties, and increased seizure risk. This condition necessitates an Individualized Education Program (IEP) or 504 plan."

Created by NERGN with funding from HRSA grant UH7MC30778. Revised 5/2024

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. 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:

- "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:

- 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

Thank you so much to Jocelyn and family for sharing their story! And thank you to the team at the Institute on Disability at UNH for assisting in collecting these stories.