Genetic Education Materials for School Success

Rubinstein-Taybi Syndrome (RTS)

Table of Contents

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.

Rubinstein-Taybi Syndrome (RTS)

At a Glance

Rubinstein-Taybi Syndrome (RTS also sometimes abbreviated RSTS) is a rare genetic condition. It is characterized by short stature, intellectual and developmental disability, distinctive facial characteristics, and other medical conditions. Both girls and boys can be affected. Most cases occur in people with no history of the disorder in their family.

Meet Malai on page 11

Common Features of RTS

- Distinctive facial features including small head, down slanting eyes, beaked nose, small mouth, low set ears
- Broad, angled thumbs and first toes
- Short stature
  - Males: 5 feet (average)
  - Females: 4 feet 10 inches (average)
- Developmental and speech delay
- Intellectual disability
- Seizures
- Vision and/or hearing problems
- Gastrointestinal issues, e.g., vomiting, diarrhea, constipation, reflux
- Behavior characteristics
  - Social and friendly in childhood
  - ADHD
  - Repetitive movements
  - Anxiety, depression, mood instability and aggression in adulthood

In Depth

Medical and Dietary Considerations

What you need to know

The list of *possible* medical problems in RTS can be extensive. However, each individual usually has only *some* of these problems. Also, the severity of any one of the possible medical conditions varies widely among individuals. Therefore, it is important to ask the parents about the unique medical issues in their child. In general, no special diet is required for RTS although a well-balanced diet is important. School age children with RTS may have multiple doctor and specialist visits to monitor medical conditions.

Medical Issues that May Be Associated with RTS

- Orthopedic Issues
    - Dislocated kneecaps
    - Scoliosis
    - Joint problems
    - Spine abnormalities (e.g., spina bifida occulta)
- Kidney problems
- Eyes/vision
    - Strabismus (crossed eyes)
    - Cataracts
    - Tear duct blockage
    - Glaucoma may be present at birth or early in life
        - May result in blindness if untreated
- Ears/hearing
    - Frequent ear infections
    - Mild hearing loss seen in ¼ of individuals.
- Heart defects
    - Most frequently patent ductus arteriosus, VSD, and ASD
    - Some require surgery
- Gastrointestinal
    - Severe constipation -can cause significant abdominal pain
    - GERD (reflux)
    - Vomiting
    - Diarrhea
- Frequent upper respiratory infections
- Seizures (~25%)
- Increased risk of developing non-cancerous and cancerous tumors
    - Certain brain tumors

    - Leukemia and/or lymphoma

What you can do

- Encourage yearly check-up and studies as needed in the child's Medical Home. Obtain copies of any updated care plans
- If vision and hearing issues are present together (two sensory systems), consult a deaf/blind specialist. Consultants for vision and hearing may help if one sensory system is affected
- If GERD (gastroesophageal reflux) is an issue, coordinate with parents
    - Identify and avoid foods that might trigger reflux
    - Avoid positions where the child's head is below their stomach, such as reading while lying down
    - Discuss the use of OTC or prescription medications with school nurse
- Follow any special diet that might be needed
- If constipation is an issue, parents may provide fiber supplements or medication
- Ensure good fluid intake
- Be aware of pain that may affect participation, performance, or behavior
- Be aware of any changes in behavior or mood that are atypical and notify the parents
- It is important to be aware of any academic changes. Contact parents when any differences are noticed

Education Supports

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

What you need to know

Individuals with RTS have a wide degree of variability in cognitive and adaptive function with many having a significant intellectual disability. Many children with RTS have delays in development. This may include delays in walking and talking. Physical and occupational therapy may help gross motor and fine motor skills. Activities that require good hand-eye control (e.g., writing, drawing, and painting) may be difficult.

Children often have difficulties with speech and articulation. Many will use sign language in combination with verbal language or other forms of communication (e.g., computer). Sensory systems such as vision and hearing may be affected. This can further impact communication.

Individuals with RTS are often described as happy, social, and loving attention. Young children with RTS often enjoy books, water play, music, and interactions with other people. They often do well in a small group and/or with a one-on-one aide. Many individuals with RTS will achieve some independence in self-care and communication.

What you can do

Teaching Strategies to Help Development

- Allow choice-making to build decision making skills
- Use routines to help learning
    - Give clear signals about the end of one activity and the beginning of a new activity
    - Use picture symbols representing activities
- Keep directions specific and brief
- Demonstrate skills to be learned
    - Use concrete objects/manipulatives along with verbal explanations
    - Use visual and auditory aids
    - Breakdown in small simple steps
- Use peer partners
- Use positive behavioral support strategies
    - Provide positive reinforcement immediately
- Have high but realistic expectations
- Small group instruction may be more beneficial to the student than whole classroom
- Present only a few stimuli or objects at a time
- Be flexible with educational goals

Strategies to Help with Short Attention Span

- Direct instruction in short periods of time
- Teach smaller chunks of activities
- Give new material slowly
- Teach in a sequential and step-by-step fashion
- Minimize distractibility
    - Keep a structured environment
    - Keep noise level down
    - Have clear expectations, routines, and rules

Strategies to Help Develop Speech and Language

- Ensure the child has a reliable way to communicate
    - They will often understand but their expressive communication may be difficult
    - This may lead the child to become frustrated
    - They may need alternative forms of communication
- Speech and language therapy and supports that are individualized to the child are helpful
    - Use a total communication approach and start as early as possible
    - Many children use sign in combination with some speech

Physical Therapy Supports

- Develop gross motor abilities
- Improve strength
- Assess modifications and adaptive equipment for school and home
    - For example, ensure desk and chair height are appropriate

Occupational Therapy Supports

- Fine motor skills
- Strength
- Dexterity
- Ensure desk and chair height are appropriate for vision and fine motor

Vision and/or Hearing Supports

- Consult a deaf-blind specialist if both hearing and vision systems are affected. Consult either a vision or hearing specialist if one system is affected

Behavioral and Sensory Support

What you need to know

Individuals with RTS may have a variety of behavioral issues. Some individuals may have a psychiatric diagnosis. Autism and/or autistic-like features are not uncommon.

Be aware that behavioral problems may be rooted in frustration over not having an adequate means of communication or there may be other triggers. Support exploration of effective means of communication for home and school and determine what triggers may be affecting their behavior.

Behavioral Issues that May Arise

- Difficulty making eye contact
- Desire for strict routine - Difficulty with change
- May react differently to sound
    - May not like loud sounds
    - May have problems in large crowds because of noise
    - May have some stimulating behaviors which are tools used for self-regulation
        - Repetitive motions
        - Hand flapping
        - Spinning
        - Rocking
- Smile often described as a grimace
- Some individuals may have psychiatric diagnoses
    - OCD, anxiety, depression

- Short attention span
- Hyperactivity
- Impulsivity
- May have abnormal response to pain

***Be aware that during adolescence other issues like anxiety or mood instability may develop**

What you can do

- Consider treatments based on individual needs
    - Behavioral supports
    - Counseling
    - Medication
- Be proactive with behavioral supports. May need to discuss involvement of behavioral/mental health professionals with the parents
    - Firm directions, rules, and clear expectations are helpful
    - May benefit from positive behavioral interventions
    - May need a functional behavioral assessment to identify causes/triggers of behaviors, functions of behaviors, and develop a plan for supporting changes
    - Ensure they have an effective means of communication
- Support children in regulating emotions and behavior
    - Teach how to self-regulate – sensory strategies may be helpful
    - Talk through expected changes
    - Consistency and routine can be helpful. Children can be easily upset with disruption in the schedule
    - Prepare them in advance for any change in schedule
    - Provide a safe area to share emotions
    - Teach and model use of words and/or pictures in sharing emotions
        - Stories may help with transitions
    - Teach, emphasize, and reinforce behaviors you want to see
    - Make sure they have an effective communication system
- Support social skills development
    - Make sure teaching strategies being used are appropriate for children who are already *socially engaged*
    - 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
    - Foster relationships and friendships with neighbors, schoolmates, and relatives so help ensure they will have friends and companions for their entire life

Physical Activity, Trips, Events

What you need to know

Physical Education:

- Exercise is important and should be encouraged; especially those exercises that build muscle mass and improve motor skills

Field Trips:

- Any change in routine may produce anxiety, fears, and/or worry
- Crowds or loud noise may produce anxiety

What you can do

Physical Education:

- Consider if adaptive physical education may be appropriate in addition to regular PE
- If reflux is a current problem, work with nurse/family to determine if any activities may trigger the reflux

Field Trips:

- Offer anticipatory guidance and preparation to prepare for a change in routine, such as a field trip
    - Use social stories and pictures to help them understand the change. The child can rehearse it alone or with others
    - Encourage use of their communication system to help them process concerns
- If a child has any sensory, hearing or vision issues, he/she may need preferred seating or other supports
- Bring any special foods or medications as needed
- Be aware of seizure protocol if the child has seizures. Make sure staff on trip are aware

School Absences and Fatigue

What you need to know

- Children with RTS may be absent due to illness and/or medical appointments
- Sleep disturbances are common and can contribute to daytime fatigue

What you can do

Absences:

- Help to make transitions in and out of school as seamless as possible

Fatigue:

- Children with RTS may be tired and require rest opportunities or breaks in their day

Emergency Planning

Develop an emergency plan if necessary, depending on the needs of individual children. For example, they may need a seizure protocol for seizures occurring during school.

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/rubinstein-taybi-syndrome/

National Organization for Rare Disorders (NORD)

NORD a large volume of information and links to helpful organizations all supporting better care for those with rare disorders. Here is their summary of RTS.
https://rarediseases.org/rare-diseases/rubinstein-taybi-syndrome/

Cincinnati Children’s Hospital

Cincinnati Children’s has a Rubinstein-Taybi Syndrome program within their Division of Developmental and Behavioral Pediatrics. Check out the information on their website.
https://www.cincinnatichildrens.org/service/r/rubinstein-taybi

University of Cincinnati Center for Excellence in Developmental Disabilities

UCCEDD has a number of great resources. Check out this downloadable pdf “Understanding Rubinstein-Taybi Syndrome: A Guide for Families and Professionals.”
https://www.ucucedd.org/wp-content/uploads/2022/08/CCHMC-Rubinstein-Taybi-Syndrome-booklet-FA_4-WEB.pdf

Info for School Nurse and Primary Care Staff

Rubenstein-Taybi Syndrome (RTS) 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 Rubenstein-Taybi Syndrome (RTS) 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/NBK1526/

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 Rubinstein Taybi Syndrome. This is a rare genetic condition that is characterized by short stature, developmental delays, speech and learning difficulties, vision and/or hearing deficits, and increased seizure risk. This condition necessitates an Individualized Education Program (IEP) or 504 plan."

Meet a young woman with RTS :

Malai - Team Player, Vibrant Athlete

"Malai is friendly and outgoing," is the first thing her mother, Molly says about Malai. "She has always been eager to learn and interact with others." Those who get the chance to interact with her have loved Malai, mostly because of her happy disposition. "We have really been blessed because of her great personality." As a child Malai was always happy. Molly reminisces that Malai never wanted to be the center of attention, but would always stay engaged with the people around her. As an adult, she has a good life in her community and has lots of friends. Molly mentions that everyone knows her everywhere they go.

Malai has always loved sports. When she was younger, she played on the same recreational soccer team as her younger brother, Mark, and their dad was the coach. Malai would play defense, and Mark would get frustrated with her because she would stand in the back and wave to people in the crowd! When she got older, she became involved in Special Olympics, where she has many friends. "When we get there, she always says hello to everyone!" remarks Molly. She has also been taking therapeutic horseback riding lessons for about 25 years, which is also a favorite sport with her brother, sister, and mother. Malai enjoys taking care of animals, including her dogs, and her mini horse named Ginger.

Malai is now 35 years old, but Molly remembers the journey through the school systems well. When Malai was about 1 year old, her parents began to question her development. They received a developmental assessment, and it was confirmed that she had delays, but they did not receive the diagnosis of Rubinstein-Taybi syndrome until later. She began to receive services while living in West Virginia, and was eventually referred to a Head Start program for 3-4 years year olds. She attended Head Start in the morning and went to therapies in the afternoon.

When Malai was 5 years old, the family moved to Massachusetts. Her parents had to bring her to a special classroom, in a special school, in a different town. They lived in Massachusetts for 9 years and then moved to Vermont. There, Malai was included in a regular classroom and was accompanied by an aid. Molly felt the school team was incredible about integrating her into everything as much as possible. They worked with her strengths and accommodated her needs. Materials were modified for Malai so she could access the curriculum. Her teachers loved working with her because she had a positive disposition and was willing to try new things. They helped her to express herself, which helped Malai to stay comfortable in the classroom. The family's stress was reduced after moving to this school and being part of a supportive team.

At school, Malai received OT and PT services for a time, but most of her services were focused on speech therapy. Malai did not start really talking until about 5 years old. Before that she learned to use sign language but was always very motivated to talk. Malai would receive speech services to help her express herself, and to help her pronounce her words more clearly. Today Malai is completely verbal but is still difficult to understand at times. Reminders to “slow down” while she’s talking really seem to help.

Molly notes that Malai has been lucky in terms of health. She wore glasses when she was young but was then told she did not need them anymore. Her teeth are crowded but have never been a problem. The family and school worked hard together to monitor her food intake and make sure she did not become overweight, something that happens often with RTS.

Malai’s school team helped transitions to go smoothly. The team anticipated some anxiety with the transition into high school. So, while Malai was in the extended school year over the summer, they had her visit the high school frequently. Becoming familiar with the school helped her to transition more easily for the school year. Molly does admit, being a professional in the field of public health and health care, she was aware of how the transition period should work and she sought out the help she needed.

Another support that proved valuable through the transition period was a curriculum the family and her school team got involved with through the University of Vermont. This curriculum is called Choosing Options and Accommodations for Children (COACH). This involved people in her life, a circle of friends, family, and educators, getting together with Malai to talk about what she sees for her future and then making a plan to get there. This started in eighth grade for Malai and her family. Molly feels this person-centered planning approach was a very positive part of planning for her whole life.

Malai also tried many jobs through the vocational services at her high school. The last job she held in high school was working at Barnes and Noble. She enjoyed being there so much that she has stayed as a part time employee there ever since. She has now been working at the same Barnes and Noble for 16 years with a job coach. She loves going in to work, and loves her co-workers, and they all love her. Malai lives at home with her parents and receives 40 hours per week of supports. She would like to live somewhere else on her own, and the family’s next adventure will be figuring out how to make that happen.

Advice for Parents:

- Meet regularly with the school team and ensure good communication.
- Find out what you can do at home to further support the services your child is receiving (i.e. speech, behavioral).
- Engage in team sports and any type of physical activities available to encourage self-esteem, more peer supports and friendships.

- Molly read the story, “Welcome to Holland” early on, which she said helped shape her views of being a parent of a child with disabilities. She recommends this poem to all parents.

Advice for Teachers:

- Communication and working on a team with the family is very important.
- Help families to know what resources and social supports are available to them.
- Ask families how you can help them- sometimes they just don’t know how to deal with problems that arise!

Thank you so much to Malai 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.