

## ***Rubinstein-Taybi Syndrome (RTS) at a Glance***

Rubinstein-Taybi Syndrome (RTS or RSTS) is a genetic condition caused in part by changes in genes, or changes on chromosome 16. Individuals with RTS typically have short stature, moderate to significant developmental delay, distinctive facial features, and broad thumbs and first toes. People with RTS usually have an intellectual disability. There is great variation in physical characteristics, medical problems, and intellectual abilities. About 1 in 100,000 to 125,000 newborns have RTS.

### **Learn More**

#### **The most prevalent findings in people with RTS**

- Differences in facial features
  - Small head
  - Downslanting eyes with highly arched eyebrows and long eyelashes
  - Thick scalp
  - Prominent beaked nose
  - Small mouth
  - Low set ears
- Thumb and first toes
  - Broad and sometimes bent
  - Found in almost all individuals with RTS
  - Surgery may be required to improve use of feet or ability to wear shoes.
  - Orthotics may be required.
- Short stature
  - Average Height
    - Males: 5 feet
    - Females: 4 feet 10 inches
  - Do not usually have a growth spurt in puberty
- Developmental delay and intellectual disability
  - In general, nonverbal IQ is higher than verbal
- Behavior
  - Social and friendly in childhood

- ADHD
- Repetitive movements
- Anxiety, depression, mood instability and aggression in adulthood

### Other Medical Issues that may be associated with RTS

- Orthopedic
  - Dislocated kneecaps
  - Scoliosis
  - Joint problems
  - Vertebral (spine) abnormalities (spina bifida occulta)
- Kidney problems or disease
- Vision (ocular)
  - 80% have some type of eye abnormality
  - Strabismus (crossed eyes)
  - Cataracts
  - Tear duct blockage
  - Glaucoma may be present at birth or early in life
    - May result in blindness if untreated
- Hearing
  - Frequent ear infections
  - Mild hearing loss seen in ¼ of individuals.
- Dental
  - Overcrowding
  - High arched palate
- Cardiac
  - Most frequently patent ductus arteriosus, VSD, and ASD (in about 1/3 of individuals)
  - Some require surgery
- Gastrointestinal
  - Severe constipation
    - May require aggressive treatment
  - GERD (reflux)
  - Vomiting
  - Diarrhea
- Frequent upper respiratory infections
- Obesity

- Anesthesia may be a problem for some people with RTS
- Seizures (~25%)
- May get fungal infections
- Tendency for scars to be raised (keloid)
- Increased risk of developing non-cancerous and cancerous tumors
  - Certain brain tumors
  - Leukemia
  - Lymphoma

## 1. Medical / Dietary Needs

### What you need to know

No special diet is required for RTS although a well-balanced diet is important, especially if a child is obese. Make sure, if applicable, that any special dietary requirements are followed.

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. Also, if reflux is a current problem, avoid positions where the child's head is below their stomach, such as reading while lying down.

They may have pain from constipation.

Children may have seizures. They may also have vision and hearing problems.

The severity of any one of the possible medical conditions varies widely between individuals. Therefore it is important to ask the parents about the medical issues in their child.

School age children with RTS may have multiple doctor and specialist visits to monitor medical conditions.

### What you can do

- Follow any special diet that might be needed
- Be aware of pain that may affect participation, performance, or behavior

- 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.
- Avoid positions that might increase reflux (GERD) *if that is a current issue or concern*. This might be, for example, reading on the floor or certain PE activities.
- A yearly check-up and studies as needed should occur in the child's Medical Home.
- 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.
- Be aware or ask parents if the child has a medical alert bracelet

## 2. Education Supports

### What you need to know

Individuals with RTS are often described as happy, social, and loving attention. Individuals with RTS have a wide degree of variability in cognitive and adaptive function with many having a significant intellectual disability. Children often have difficulties with speech and articulation. Many will use sign language in combination with verbal language or other forms of communication (i.e. computer).

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. They often participate in supported work situations as adults.

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 (i.e. writing, drawing, and painting) may be difficult.

Sensory systems may be affected, such as vision and hearing.

Due to short stature, some accommodations may need to be made.

## 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.

### Teaching 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 sequentially and step-by-step fashion
- Minimize distractibility
  - Keep a structured environment
  - Keep noise level down
  - Have clear expectations, routines, and rules

### To help develop speech and language

- Ensure the child has a reliable way to communicate

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- 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 total communication approach and start as early as possible.
  - Many use sign in combination with some speech

### Physical Therapy can help to

- 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 can help to develop

- 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.

## 3. Behavioral and Sensory Support

Individuals with RTS may have a variety of behavioral issues. Some individuals may have a psychiatric diagnosis; some may have autism and/or autistic-like features. It is important to treat children who have RTS syndrome as an individual and provide the supports they need to be successful.

Behavioral problems may be rooted in their frustration over not having an adequate means of communication or there may be other triggers. Adults need to support exploration of effective means of communication for home and school and determine what triggers may be affecting their behavior.

## What you need to know

### 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 self-stimulating behaviors
    - Repetitive motions
    - Hand flapping
    - Spinning
    - Rocking
- Smile often described as a grimace
- Some individuals may have psychiatric diagnoses
  - OCD
  - Anxiety
  - Depression
  - Consider need for behavioral therapy or medication
- Short attention span
- Hyperactivity
- Impulsivity
- May have abnormal response to pain

### During adolescence, other issues may develop

- Anxiety
- Mood instability

## 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 and/ or medications 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
- Many children have difficulty regulating emotions and behavior. This is especially true when handling unplanned changes.
  - 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.
  - Teach how to regulate own body – sensory strategies may be helpful.
  - Foster relationships and friendships with neighbors, schoolmates, and relatives so help ensure they will have friends and companions for their entire life.

## 4. 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.

- 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
- Be aware of the seizure protocol if the child has seizures.

### **Field Trips:**

- Any change in routine may produce anxiety, fears, and/or worry.
- Offer anticipatory guidance and preparation to prepare for a change in routine such as a field trip.
- Create a picture story about the upcoming event. The child can rehearse it alone or with others.
- If a child has any sensory, hearing or vision issues, he/she may need preferred seating.
- Crowds or loud noise may produce anxiety
- Be aware of seizure protocol if the child has seizures

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**

- Offer anticipatory guidance and preparation to prepare for a change in routine, such as a field trip.
- Offer supports as needed for vision and hearing issues.
- 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**

### **What you need to know**

#### **Absences:**

- Children with RTS may be absent due to illness and/or medical appointment

#### **Fatigue:**

- Sleep disturbances may be common

## **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

## **6. Emergency Planning**

### **What you need to know**

- 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.

## **7. Resources**

### **Rubinstein-Taybi Syndrome Support Group**

*The parent support page with information for parents and health professionals*  
<http://rubinstein-taybi.com/>

### **Genetics Home Reference**

Consumer-friendly information about human genetics from the U.S. National Library of Medicine  
<http://ghr.nlm.nih.gov/condition/rubinstein-taybi-syndrome>

### **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>