Rubinstein-Taybi Syndrome (RTS)  
For Healthcare Providers

This is a customized health care provider version of our website. Please visit the main website to find more comprehensive information for families and schools ([www.gemssforschools.org](http://www.gemssforschools.org)).

Physical characteristics and/or symptoms

*Note: not all people with RTS will have all of these features.*

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
- Short stature
  - Average Height
    - Males: 5 feet
    - Females: 4 feet 10 inches
- Developmental delay and intellectual disability
- Behavior
  - 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 abnormalities

Kidney problems or disease

Vision
- Strabismus
- Cataracts
- Tear duct blockage
- Glaucoma may be present at birth or early in life

Hearing
- Frequent otitis media
- Mild hearing loss

Dental
- Talon cusps, an accessory cusp-like structure on the lingual side of the tooth, usually occurring on the maxillary incisors of the permanent dentition
- Overcrowding
- Narrow palate

Cardiac
- Approximately one third have cardiac malformations
- Most frequently patent ductus arteriosus, VSD, and ASD

Gastrointestinal
- Severe constipation
- GERD
- Vomiting/Diarrhea

Frequent upper respiratory infections

Obesity
- Obstructive sleep apnea

Anesthesia may be a problem for some people with RTS

Seizures

Increased risk of developing non-cancerous and cancerous tumors
- Certain brain tumors
- Leukemia
- Lymphoma

Recommended Routine Surveillance
• Monitor of growth and feeding
• Routine monitoring for cardiac and renal anomalies
• Annual hearing and eye evaluations
• Routine dental care

Emergency Protocols

There are no specific emergency protocols for this particular condition as it is not typically associated with episodes of sudden and serious medical decompensation.
• Emergencies should be handled as with any child.
• If seizures are present, the following seizure action plan may be useful:

Specialists Who May Be Involved

Follow up is need on a case-by-case basis. A multidisciplinary team approach to best meet the child’s individual needs is recommended.

• Cardiologist
  o Structural heart defects
• Dental
  o Talon cusps, an accessory cusp-like structure on the lingual side of the tooth, usually occurring on the maxillary incisors of the permanent dentition
• Developmental evaluation
  o Speech therapy with emphasis on nonverbal methods of communication
  o Physical therapy
  o Occupational therapy
• ENT
  o Hearing loss
  o Obstructive sleep apnea
• Gastroenterologist:
  o Significant feeding problems may occur
  o Gastroesophageal reflex
  o Constipation
• Geneticist / Genetic Counselor:
  o Diagnosis
  o Coordination of care
  o Genetic risk for family

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Clinical trials

• Hematology/Oncology
  o Treatment for leukemia or other malignancies
• Neurology
  o Monitor seizures
• Nutritionist
  o Weight control
• Orthopedists
  o Monitor gait impairment
  o Scoliosis
  o Muscular hypotonia
• Ophthalmology
  o Strabismus
  o Cataracts

Sample Forms

• Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

  My patient______________________ has been diagnosed with Rubeinstein-Taybi syndrome. Distinct facial features, broad thumbs and toes, short stature and moderate and to severe intellectual disability characterize Rubenstein-Taybi syndrome. Medical complications with Rubenstein-Taybi syndrome include management of seizures, growth and feeding difficulties, cardiac defects, gastrointestinal reflux, and constipation, Because of these, ______ needs the following accommodations.

Seven Important Aspects of School Life

“Rubenstein-Taybi Syndrome at a Glance” will help you talk with parents and schools about:

• Medical / Dietary Needs
• Education Supports
• Behavior & Sensory Supports
• Physical Activity, Trips, Events
• School Absences & Fatigue
• Emergency Planning
• Transitions

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Resources

Genetic Home Reference

GeneReviews
https://www.ncbi.nlm.nih.gov/books/NBK1526/