

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).

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

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- 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

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- 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:
https://www.aap.org/en-us/Documents/Seizure_Action_Plan_for%20School.pdf

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
 - Structural heart defects
- 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
- Developmental evaluation
 - Speech therapy with emphasis on nonverbal methods of communication
 - Physical therapy
 - Occupational therapy
- ENT
 - Hearing loss
 - Obstructive sleep apnea
- Gastroenterologist:
 - Significant feeding problems may occur
 - Gastroesophageal reflex
 - Constipation
- Geneticist / Genetic Counselor:
 - Diagnosis
 - Coordination of care
 - Genetic risk for family

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

Sample Forms

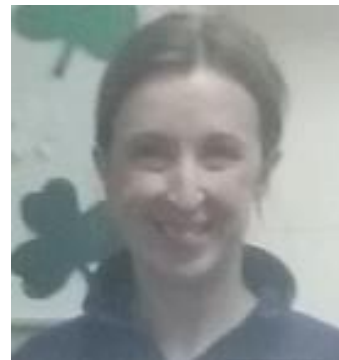
- Sample paragraph to be used for Letters of Medical Necessity or Letters to the school:

My patient _____ has been diagnosed with Rubenstein-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

<https://ghr.nlm.nih.gov/condition/rubinstein-taybi-syndrome>

GeneReviews

<https://www.ncbi.nlm.nih.gov/books/NBK1526/>