



The Arc Answers: Rubinstein-Taybi syndrome

How many people have Rubinstein-Taybi syndrome?

Estimates vary, but approximately 1 in every 100,000-125,000 individuals are born with Rubinstein-Taybi syndrome. It occurs in all ethnicities and with equal frequency in males and females.

What causes Rubinstein-Taybi syndrome?

Researchers are still trying to determine the cause or causes of Rubinstein-Taybi syndrome. In about half of all cases, there appears to be randomly mutated or deleted material from one of two specific genes on chromosome 16. However, in the other half of all cases, these genes do not appear to be affected, even though children share the same distinctive physical characteristics and symptoms. Nobody knows why these mutations occur. In most cases, a person with Rubinstein-Taybi syndrome is the first person in the family to have the syndrome.

What are the primary characteristics of Rubinstein-Taybi syndrome?

People with Rubinstein-Taybi syndrome have distinctive physical characteristics, including short stature, broad thumbs and big toes, widely spaced eyes, a beaked nose, small jaw, and low set ears. Each person has a unique combination of characteristics.

Behavioral characteristics may include: a short attention span, impulsivity, obsessive-compulsive or mood disorders, and withdrawal. People with Rubinstein-Taybi syndrome also have some level of cognitive delay that differs with each person.

What are common medical concerns?

Children and adults have unique medical concerns; no person will have all of the concerns listed. Parents may need to seek help for their children regarding: feeding difficulties, dental problems, respiratory infections, kidney issues, ear and eye infections, cardiac issues (in about 30 percent of children), tear duct obstruction, sometimes diarrhea but usually chronic constipation, undescended testicles in males, and skeletal issues. In addition, some individuals do not tolerate anesthesia well.

What interventions have been proven helpful for people with Rubinstein-Taybi syndrome?

As most children with Rubinstein-Taybi syndrome experience delays with speech or may remain nonverbal, speech therapy, sign language, or augmentative technology can help facilitate communication. Children who need help developing gross and fine motor skills may respectively benefit from physical and occupational therapies. Upon entering the school system, special education can help tailor an educational program to a child's developmental level. As each child is unique, a personalized plan should be developed in conjunction with professionals to best help a child reach his or her full potential.

Are there any sources of financial support for families?

The Special Friends Foundation handles requests for financial assistance to be applied to medical expenses on a case-by-case basis. More information can be found on their website:

<http://www.specialfriends.org/Financial.html>

Where can I find other resources?

Rubinstein-Taybi Association: <http://www.rubinstein-taybi.org/>

RTS and anesthesia: <http://www.rubinstein-taybi.org/bluebook/anesthesia.html>

Special Friends Foundation: <http://www.specialfriends.org/index.html>

US National Library of Medicine: <http://ghr.nlm.nih.gov/condition=rubinsteintaybisynndrome>

US National Institutes of Health: <http://www.nlm.nih.gov/medlineplus/ency/article/001249.htm>

Madison's Foundation – Rare Disease database:

http://www.madisonsfoundation.org/index.php/component?option=com_mpower/Itemid,49/diseaseID,590/

The Arc of NC
343 East Six Forks Rd. Ste.320
Raleigh, NC 27609
1-800-662-8706
www.arcnc.org