

Rubinstein-Taybi Syndrome

WHAT IS RUBINSTEIN-TAYBI SYNDROME?

Rubinstein-Taybi Syndrome (RTS) is a genetic multi-system disorder characterized by facial abnormalities, broad thumbs and great toes, and developmental disability. It was first described in 1957 and was identified as a recognizable syndrome in 1963 by Dr. Jack Rubinstein and Dr. Hooshang Taybi.

HOW IS RTS DIAGNOSED?

The diagnosis of RTS can be made following a complete clinical exam including X-rays of the hands, a CT-scan or MRI, an EEG to evaluate the electrical activity of the brain, and an EKG or echocardiogram. Furthermore, RTS may be confirmed by genetic testing for a deletion or mutation in chromosome 16p, although not all patients carry a mutation in this area. While some children with more severe characteristics are diagnosed at birth, those more mildly affected may not be diagnosed until adolescence.

WHAT ARE THE OCULAR SIGNS OF RTS?

The ocular signs of RTS include downward slanting of the eyes, widely spaced eyes, [strabismus](#) (eye misalignment), [ptosis](#) (droopy eyelid), high arched eyebrows, and frequent eye infections due to blockage of the tear drainage ducts. There is also a less-common association with congenital glaucoma, characterized by excessive tearing of the eyes, larger than normal eyes, and/or cloudy corneas.

IS RTS HEREDITARY?

In many cases, RTS is caused by a deletion or mutation in the short arm (p) of chromosome 16. RTS is found equally in both males and females and is rare, occurring in 1 out of every 100,000 to 300,000 live births. In most cases, the occurrence of RTS is random with no other family members with the syndrome. There is, however, an increase in the number of cases being reported each year.

WHAT NONOCULAR PROBLEMS DO PATIENTS WITH RTS HAVE?

There are many characteristics associated with RTS. Not every individual has all the characteristics, however, the following is a list of the reported traits:

- Broad thumbs and/or toes (sometimes angulated)
- Mental retardation (from mild to severe)
- Beaked nose
- Short stature (delayed bone age)
- Broad nasal bridge
- Malformed ears
- High arched palate
- Extra fold of skin on either side of the nose
- Small head (microcephaly)
- Small lower jaw
- Flat red birthmark on forehead
- Hyperextensible joints
- Small tilted pelvis
- Excessive hairiness
- Undescended testicles
- Feeding difficulties
- Respiratory infection
- Cardiac anomalies
- Vertebral abnormalities
- Gastroesophageal reflux and vomiting
- Kidney abnormalities
- Orthopedic problems

WHAT ARE THE TREATMENT OPTIONS FOR RUBINSTEIN-TAYBI SYNDROME?

The symptoms of RTS range from mild to severe, therefore treatment programs should be developed on an individual basis. Early intervention with an emphasis on speech therapy is advised. If verbal communication skills are severely delayed, sign language as an alternate/additional form of communication is taught. If skeletal abnormalities are problematic, physical and occupational therapy may be recommended.

WHERE CAN I GET MORE INFORMATION ABOUT RUBINSTEIN-TAYBI SYNDROME?



- Further information can be found at specialfriends.org. There is a Facebook group at www.facebook.com/RTSCF.

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