Broad Thumb-Hallux Syndrome
(Rubinstein-Taybi Syndrome)

What is RTS and its Symptoms
Genetic disorder including short stature, mental retardation (moderate to severe), eyes slant downward and are far apart, broad thumbs and big toes. Babies that are born with RTS are normal length but stop growing as they get older.

History and Cause
Rubinstein-Taybi was first described in 1963 by Dr. Jack Rubinstein and Dr. Hooshang Taybi. This syndrome is inherited in an autosomal dominant manner. Some cases are caused by mutation in a gene called CREBBP (located in chromosome 16). CREB binding protein is necessary for cell growth in particular cell division. It is inheritable because at this point you have a good copy and a bad copy of chromosome 16 so there is a 50/50 chance of passing it to its siblings. It is sporadic in the way that mutation occurs randomly.

Test and Diagnosis
There are no specific tests to diagnose RTS. Genetic testing for the GREPPT gene can only confirm the diagnosis. X-Rays, echocardiograms, and electrocardiograms only show current or future symptoms. Examination and testing may reveal absent or extra kidneys, cataracts or other eye defects.

Treatment
Even though there is no cure for RTS, there are many techniques that can help manage the condition:
- Special Education Programs- available for them at public schools
- Speech Therapy-for children with speech problems
- Physical and Occupational Therapy-if bone problems are experienced
- Nutritional Counseling-Patient can overeat or can also be underweight
- Dentist Visits-regular visits due to crowding of teeth
- Cardiologist/Nephrologists-If heart or kidney problems are experienced
- Behavior specialist/Child psychiatrist-for behavior problems

Test have found that this disorder occurs in 1 in 130,000 births. Genetic counseling is advised for couples with a family history of this disease who are planning a pregnancy. It is assumed that individuals with Rubinstein-Taybi syndrome are capable of having children.

Citations