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Rubinstein–Taybi Syndrome

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Introduction

- Dr. Jack Rubinstein and Dr. Hooshang Taybi who recognized this pattern in seven unrelated children first described the condition in 1963. It occurs in an estimated 1 in 125000-300000 births.

Rubinstein–Taybi syndrome (RTS), also known as broad thumb-hallux syndrome or Rubinstein syndrome, is a condition characterized by short stature, moderate to severe learning difficulties, distinctive facial features, and broad thumbs and first toes.

Genetics

- Rubinstein–Taybi syndrome is a micro deletion syndrome involving chromosomal segment 16p13.3 and is characterized by mutations in the CREBBP gene. The CREBBP gene makes a protein that helps control the activity of many other genes. The protein, called CREB-binding protein, plays an important role in regulating cell growth and division and is essential for normal fetal development. If one copy of the CREBBP gene is deleted or mutated, cells make only half of the normal amount of CREB binding protein. A reduction in the amount of this protein disrupts normal development before and after birth, leading to the signs and symptoms of Rubinstein–Taybi syndrome
- Mutations in the EP300 gene are responsible for a small percentage of cases of Rubinstein–Taybi syndrome.

Inheritance

- In almost all cases, there are no other family members with the Rubinstein-Taybi syndrome. Based on the small number of recurrences in families, the risk to have a second child with Rubinstein-Taybi syndrome is probably less than 1%. However, the risk for a person with Rubinstein-Taybi syndrome to have an affected child is probably much higher, possibly as high as 50%.
- At the present time, there is no way to test a baby prenatally (before birth) to see if he/she has the condition.

Medical Problems

A variety of medical problems can be seen in children with Rubinstein-Taybi syndrome.

However, it is unlikely that an individual child would have all of these difficulties.

- General features: unusual facies, Mental disability and poor weight gain (due to feeding problems like swallowing difficulty or vomiting)
- Musculoskeletal features: Broad thumb and first toes (almost all patients). Dislocated patella and scoliosis, pectus excavatum
- Ophthalmic features: Eye abnormality (80%) –down slanting of palpebral fissure, colobomas,ptosis, strabismus, cataract, glaucoma or tear duct obstruction. Thick arched eyebrows with long eyelashes can also be seen.
- Cardiovascular features: Congenital heart defects (35-40%) most commonly being PDA. Others include pulmonary stenosis, aortic coarctation, VSD's and ASD'S.
  Otorhinolaryngologicfeatures: Malformed ears. Ear infections very frequently with mild degree of hearing loss.
- Respiratory features: recurrent upper respiratory tract infections and sleep apnoea
- Orthodontic features: Dental problems due to overcrowding of teeth and increased frequency of cavities. High arched palate. Hypoplastic maxilla.
- Gastrointestinal features: Severe Constipation
- Genitourinary features: cryptorchidism, or hypospadias may be present. In some cases they may have Hypoplastic or absent kidney, repeated UTI's, kidney stones, hydronep-hrosis or duplicated ureter can also be present.
- Others features includes increased incidence of keloid formation and increased suscep-tibility to develop certain malignancies.

Life of a Rubinstein Taybi Syndromic Child

- **Growth:** Most children with Rubinstein-Taybi syndrome are of average weight and length at birth. However, within the first few months, growth falls well below average. They are short throughout life and do not seem to have the usual growth spurt around puberty. The average adult height in males is 5', while in females it is 4'10" (but this is variable).
- **Puberty & Fertility:** The onset of puberty occurs at approximately 12 years of age, which is similar to the general population. Young women began to have menstrual cycles at the usual age as well. According to studies, individuals with Rubinstein-Taybi syndrome are physically capable of having children.
- **Development:** All individuals with Rubinstein Taybi syndrome have developmental delay, however the degree of limitation is variable. The average IQ (intelligence quotient) ranges from 30-79 (normal range is 85-115). Speech problems are present in about 90% of patients.
- **Personality:** Each child with Rubinstein-Taybi syndrome is a unique individual with his/her own personality. However, in general, their families as loving, friendly, and happy as opposed to irritable or unpleasant describe children with Rubinstein-Taybi syndrome.
- **Behaviour:** Sometimes behaviour in a handicapped child can be the greatest concern of his/her parents. Some children with Rubinstein-Taybi syndrome engage in occasional self-stimulatory behaviour such as rocking, spinning, and hand flapping. A short attention span was described in 90% of patients. Many of the children seem to dislike loud sounds or do not tolerate crowds because of noise.

Safety of Immunisation

The overall complication rate was only 5%, which is no greater than the general population rate. Reactions consisted primarily of low-grade fever and irritability.

Thus, there seems to be no reason to withhold immunizations in these children. As in all children, immunizations are important in order to prevent serious infectious diseases.

Management

There is no specific treatment for RTS.

Surgery to repair the bones in the thumbs or toes can sometimes improve grasp or relive discomfort