

Rubinstein Taybi Syndrome

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Introduction

Rubinstein-Taybi is a rare genetic syndrome¹ characterised by distinctive facies, microcephaly, mental retardation, broad thumb and great toe with other abnormalities. Diagnosis is mainly clinical. Incidence is 1:100000-125000 at birth². In 1963 this syndrome was first described by Rubinstein and Taybi¹.

The Case

A two and a half year old Muslim female child, born out of nonconsanguineous marriage, presented with complaints of inability to walk and speak. She is the first child of the parents. She could only stand with support, produce some incomprehensible sounds. There is past history of frequent cough and cold with ear discharge, recurrent purulent eye discharge and frequent watering from both eyes. During infancy the baby had difficulty in feeding. On examination, the child was happy and easy going, had microcephaly (OFC: 42cm, expected: 47.5cm), short stature (height: 70cm, expected: 92cm), mental retardation. Typical facial features included broad forehead, frontal hair up-sweep, pointed beaked nose, highly arched eyebrows, micrognathia, high arched palate, low-set deformed ears, bilateral pits on the posterior aspect of helix, short neck, downward slanting of palpebral fissures. Examination of hands and feet revealed broad thumbs and great toes with radial deviation of thumbs, post axial polydactyly in lower limbs, persistent fetal fingertip pads, fifth finger clinodactyly, syndactyly between great toe and 2nd toe of left lower limb. Echocardiographies, USG abdomen, EEG, X-ray spine, ophthalmological examination were normal. There was no family history of similar disease and parents were apparently normal. Among the

Abstract

Rubinstein Taybi is a rare genetic syndrome with characteristic facial features, broad thumb and toes, mental retardation. Diagnosis is mainly clinical. Here we report a 2 and ½ year old female child with typical facial features and other anomalies characteristics of Rubinstein Taybi syndrome. Among the different features described-polydactyly, syndactyly, micrognathia are occasional abnormalities to be found in this syndrome. Bilateral pits on posterior aspect of helix is also a feature described in very few literatures.

different features described-polydactyly, syndactyly, micrognathia are occasional abnormalities to be found in this syndrome³. Bilateral pits on posterior aspect of helix is also a feature described in very few literatures.



Fig 1: Showing patient having typical facies, post axial polydactyly, broad and radially deviated thumb. (The mark in the middle of forehead is not a skin lesion but a religious tika)

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Discussion

Diagnosis of RTS is still clinical⁴. Majority of cases are sporadic. It can also be caused by microdeletion of 16p13.3 or by mutation in either CRE B-binding protein (CBP) or E1A-binding protein (p300). About 4-25% cases are due to microdeletion detectable by FISH³. However Clinical features are usually same in those with or without deletion with possible exceptions of microcephaly, angulation of fingers and partial duplication of halluces which are more common in those with microdeletion. Cyclic-AMC-regulated enhancer (CRE) binding protein, or CREB binding protein, is generally referred to as CBP. CBP has a homolog, *p300*, located at chromosome 22q13; both are similar in structure and function⁵: both act as transcriptional co-activators and act also as potent histone acetyl transferases by making the DNA accessible to transcription factors⁶. They are mediators of signaling pathways and participants in basic cellular functions such as DNA repair, cell growth, cell differentiation, apoptosis, and tumor suppression.

Continuing research eventually led to the demonstration of mutations in the causative *CBP* gene. Combining the results of larger studies⁷, a *CBP* mutation was found in 63 of 155 patients (41%). Subsequent studies showed mutations in *p300* in a limited number of individuals⁷. The true frequency of *p300* mutations remains uncertain. At present it remains uncertain whether there is yet another cause of Rubinstein-Taybi syndrome.

Conclusion

An early diagnosis is critical both for the parental information and for management of the medical problems. Adequate counseling of parent include first properly formulated information regarding the disease

itself. Regarding recurrence, in sibling recurrence risk 0.1 percent⁷. These children can reproduce, and recurrence risk in their offsprings is 50 percent⁸. These patient have increased risk of certain tumor mainly meningioma, brain tumors, and leukemia. Life expectancy seems to be normal. As these patients are mainly asymptomatic, most of them remain undiagnosed.

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