The Professional Medical Journal
www.theprofesional.com

DOI: 10.17957/TPMJ/16.3478

CASE REPORT PROF-3478

RUBINSTEIN TAYBI SYNDROME;
A VERY RARE CONDITION

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Article received on:
06/06/2016
Accepted for publication:
30/06/2016
Received after proof reading:
04/07/2016

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ABSTRACT... Rubinstein Taybi Syndrome (RTS) was first described in 1963 by Rubinstein and Taybi. The characteristic features of this syndrome include broad thumbs and toes, facial abnormalities like hypertelorism, beaked nose, micrognathia, microcephaly and mental retardation. Cardiac, renal, ophthalmological and various orthopedic problems can also occur. Prevalence in the general population is approximately 1 case per 300,000 persons and is as high as 1 case per 10,000 live births. There is no definite inheritance pattern so far and recurrence is very unlikely. In some patients, multiple chromosomal anomalies have been described. We report here a case of Rubinstein Taybi syndrome in an 18 months old girl presented with typical features which is the first case reported in our population.

Key words: Rubinstein Taybi syndrome, Chromosome, Mental retardation, CBP gene.

Article Citation: Khan F, Cheema TA, Tahir M. Rubinstein taybi syndrome; a very rare condition. Professional Med J 2016;23(7):883-886. **DOI:** 10.17957/TPMJ/16.3478

INTRODUCTION

Rubinstein Taybi syndrome (RTS) or broad thumb- hallux syndrome is a multiple congenital anomaly syndrome characterized by broad thumbs, big and broad toes, short stature and craniofacial anomalies like hypertelorism, beaked nose, micrognathia, microcephaly, and mental retardation.[1] High arched, narrow palate, cleft uvula, cleft palate and cleft upper lip can also be part of the syndrome.[2] Cardiac, renal, ophthalmological, various orthopedic problems and childhood tumors can also occur.[2] Prevalence in the general population is approximately 1 case per 300,000 persons and is as high as 1 case per 10,000 live births[3]. In mental retardation clinics, RTS is present in approximately 1 in 600 patient's seen[4]. There is no definite inheritance pattern so far and recurrence is very unlikely. The cause of RTS is still unclear. Recently, RTS was shown to be associated with disruption of the binding protein for cyclic adenosine monophosphate–response element binding protein (CBP), either by gross chromosomal rearrangements or by point mutations.[3] The administration of GA in RTS patients is usually complicated because of craniofacial and cardiac abnormalities and gastro esophageal reflux.[3]

CASE REPORT

An 18 months old girl was referred to National Orthopedic Hospital, Bahawalpur for evaluation of congenital deformity of her both hands and feet. We enquired the history from parents accompanying the patients. There was no history of family consanguinity. It was found that the girl was born at term by caesarian section and her perinatal history was unremarkable. Weight of the girl at birth was 3.5 kg. Her developmental milestones were late and the parents told that the girl is slow in learning things of daily living. The mother of the baby was a 28 years old primipara and denied any other relevant obstetric history. Her height was 60cm and Occipito-frontal circumference (OFC) was 36cm. She was overweight and her present weight was 9 kg. Physical examination of the girl revealed nearly all the characteristic features of RTS including broad thumbs bilaterally, big and broad toes, short stature (Height 60cm), microcephaly (OFC 36cm), hypertelorism, ptosis, short and broad beaked nose and micrognathia. On intraoral examination, she was having high arched narrow-grooved palate with crowded teeth and submucus cleft soft palate. Routine blood investigations were normal. Evaluation of the radiographs of both

hands and feet revealed deviated broad thumbs and big toes bilaterally. There was deviation of > 70 degrees at thumbs interphalangeal (IP) and > 30 degrees at big toes IP joints. The opinions of cardiologist, ophthalmologist and gastroenterologist were sought. Sonographic and echocardiographic evaluation of the neonate did not reveal any significant finding.

(a) (b) (c) (d)

Figure-1. An 18 – months old child having broad and deviated thumbs, big and broad toes.

Figure 2. Same patient with craniofacial anomalies like hypertelorism, beaked nose, micrognathia, microcephaly, high arched, narrow and cleft palate.

DISCUSSION

Rubinstein Taybi syndrome (RTS) is the classical recurrent- pattern multiple congenital anomaly syndromes. In the year 1957, Rubinstein evaluated a 3.5 year-old girl with atypical facial appearances and broad thumbs and great toes[5]. Michail et al from Athens published a case report in a French orthopedic journal in the same year[4]. Since then more than 700 cases have been reported world-wide.

This 18 months old girl in our case has got typical manifestations of RTS. Our patient was characterized by short stature, broad deviated thumbs and big toes, hypertelorism, ptosis, broad beaked nose, small jaw, and low set ears. In RTS, various behavioral problems like short attention span, impulsivity, obsessive-compulsive or mood disorders can occur. Some patients also have some level of cognitive delay that differs with each person.[2] Abnormalities of eye and renal system are frequent, as are various orthopedic problems serious enough to delay motor milestones and cause gait impairment in later life.[4] Orthopedic abnormalities have made possible in diagnosing an individual with probable RTS. There are numerous reports of various types of childhood tumors as well.[2] Our case showed some developmental delay in attaining milestones and was overweight and short stature.

The cause of RTS is still unclear. Recently, RTS was shown to be associated with disruption of the binding protein for cyclic adenosine monophosphate–response element binding protein (CBP), either by gross chromosomal rearrangements or by point mutations.[3] Hennekham et al. found a micro deletion in 25% of patients at 16 p13.3 region in some patients and suggested that deletion is the most probable

cause of syndrome.[6] Transmission from parent-to-child has been reported in one case; furthermore a mother and daughter, both of whom appear to be affected with RTS strongly suggest either autosomal or X-linked dominant transmission.[4] In our case, parents were unrelated and she was the only child delivered by caesarian section and there was no family history of RTS.

In the literature, mostly there are case reports on RTS. One large series is described by Cantani and Gagliesi in 1998 in which they analyzed 732 cases of RTS.[4] They identified the typical cases and review the symptom and signs of this syndrome by meta-analysis of 17 papers. Bellini and Boniolo report further 11 cases with age-range 1-13 years.[7] Battaglia and Ferrari report 6 cases of this syndrome and they investigate particularly the cognitive and psychological profiles of these patients.[8] All these studies have shown that broad thumbs and toes and typical facies are the most consisting features of RTS. In our case, X-rays of both hands and feet revealed broad deviated thumbs and big toes. Other specific investigations like complete blood examination, abdominopelvic ultrasound, echocardiography and CT scan brain should be done to rule out associated cardiac, renal and brain problems. We did all these investigations and were unremarkable. Ophthalmological problems like refractive errors, nasolacrimal duct obstruction, strabismus and ptosis can occur in RTS.[4] We observed hypertelorism and grade 2 ptosis in our case. These patients have also got high arched and narrow palates with crowded teeth causing problems in feeding and maintaining oral hygiene especially in illiterate family.[3] The parents of our patient were educated and her mother was a school teacher and she maintained her oral hygiene quite well.

Potential problems can occur in RTS while administering the endotracheal tube during general anesthesia because of the limited mouth opening, the high-arched palate, and the micrognathia in these patients.[3] In addition, the history of recurrent respiratory infections and the evidence for gastro esophageal reflux in RTS required extreme caution. In addition to the anesthesia-related risks, other risks in RTS patients have been reported. These include supraventricular and ventricular dysrhythmias, hypotonia, seizures, respiratory and cardiac failure.[4]

In Pakistan, there is no case of RTS described so far. We think that by reporting this case, the clinician should bring RTS in their diagnosis if they come across such patient with broad deviated thumbs and big toes and typical facies. When a diagnosis of RTS is made, ophthalmologic, renal and orthopedic evaluations should all be considered. Precautionary measures should be adopted while administering the general anesthesia; anesthesiologists should be informed that this patient has RTS and information offered.

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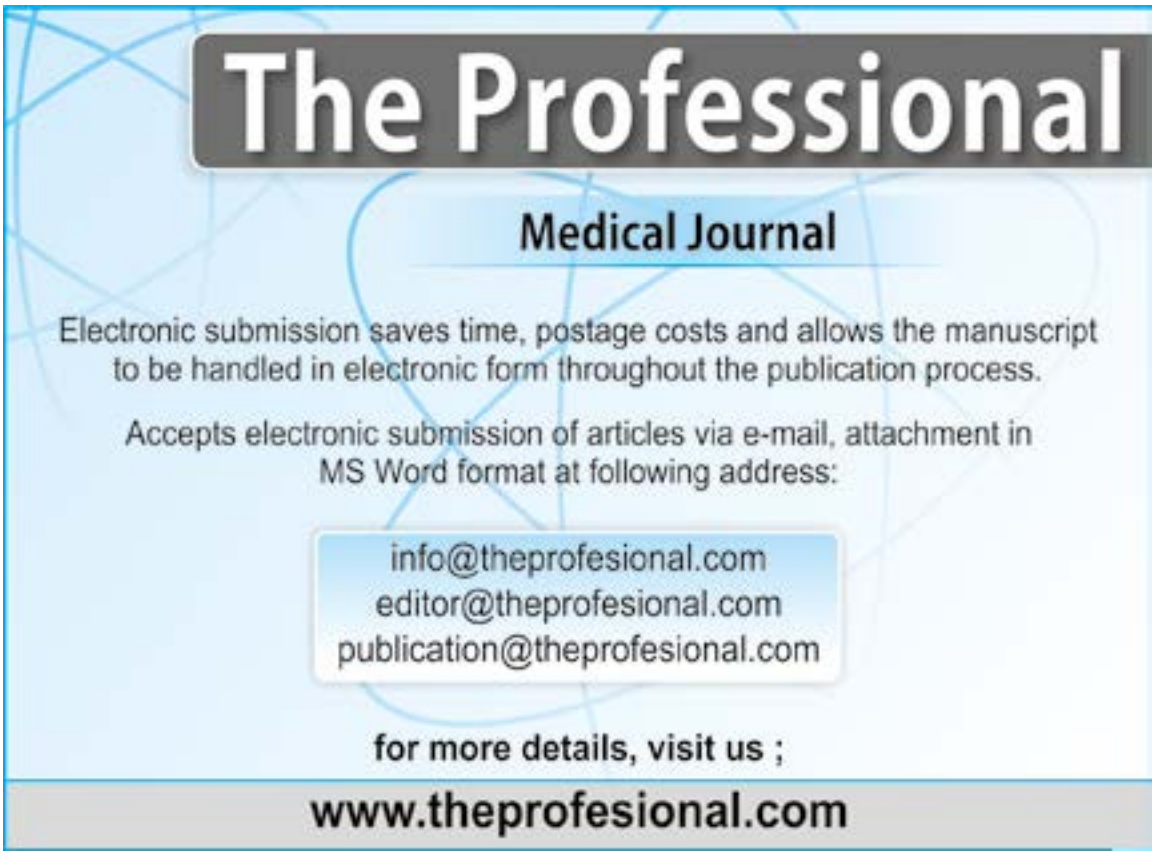

AUTHORSHIP AND CONTRIBUTION DECLARATION

Sr. #	Author-s Full Name	Contribution to the paper	Author=s Signature
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2	Dr. Tahseen Ahmed Cheema	Literature Review, Proof Reading	
3	Dr. Muhammad Tahir	Introduction, Analysis	