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Case report

An unusual presentation of Rubinstein-Taybi Syndrome with bilateral postaxial polydactyly

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Abstract

Rubinstein-Taybi Syndrome (RSTS) is a rare multiple congenital anomaly syndrome. Only 250 cases have been described in medical literature. We hereby present a 10-month-old male child with characteristic facial features and hand and feet anomalies. The characteristic features of hands and feet typically described are broad thumbs and halluces and clinodactyly of the 5th finger, whereas polydactyly is a rarely reported feature in this syndrome. This case promotes awareness regarding this syndrome and emphasises rarely reported features that should raise high degree of suspicion in a child presenting with multiple congenital anomalies and have a great importance in diagnosis of a genetic syndrome like RSTS. Early detection is essential for prevention of morbidity, mortality and disability.

Keywords

Rubinstein-Taybi Syndrome, polydactyly, multiple congenital anomalies.

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Introduction

Rubinstein-Taybi Syndrome (RSTS) (OMIM Entry #180849) is a quite rare congenital syndrome, characterised by short stature, intellectual disability, distinctive facial features and broad and angulated thumbs and halluces. It was first described by Rubinstein and Taybi in 1963 [1]. The

incidence of RSTS is 1 in 100,000 to 125,000 live births [2]. Only 250 cases have been described in medical literature [3]. The characteristic craniofacial features include down-slanted palpebral fissures, low hanging columella, high palate, grimacing smile, and talon cusps [4]. Other variable findings are neurological abnormalities [5] including posterior fossa abnormalities, cervical cord compression, eye abnormalities including retinal dysfunction on electroretinography (ERG) [6] and strabismus, coloboma, cataract, congenital heart defects, renal abnormalities, and cryptorchidism [4]. Orthopedic issues include dislocated patellas, lax joints, spine curvatures, Legg-Calve-Perthes disease, slipped capital femoral epiphysis, and cervical vertebral abnormalities [7]. These children are also predisposed to various malignancies including meningioma, rhabdomyosarcoma, pheochromocytoma, leukemia and also other tumors [8].

Case presentation

A 10-month-old boy, second in birth order, born of non-consanguineous marriage, with uneventful antenatal and perinatal history, presented to us for multiple congenital anomalies. There was no family history of intellectual disability or similar features. On clinical examination his anthropometry parameters were as follows: weight, 6.5 kg (between -2 and -3 SD); length, 64 cm (< -3 SD); head circumference, 40 cm (< -3 SD). His characteristic craniofacial features (**Fig. 1**) included microcephaly with flat occipit, frontal bossing, medial flaring of eyebrows, prominent pinched nose, nasal septum extending beyond alae nasi, high arched palate, micrognathia, small mouth and anteriorly rotated prominent ears. Examination of hands and feet revealed broad thumbs with radial angulation (**Fig. 2A**), persistent finger pads (**Fig. 2B**), camptodactyly of 3rd, 4th and 5th fingers (**Fig. 2B**), broad halluces with bilateral postaxial polydactyly (**Fig. 3**). He also had undescended testes. History of constipation was present since birth. The child had global developmental delay with developmental age of 5 months (development quotient: 50%).

Routine blood investigations, including haemogram, coagulation profile, and biochemical profile, were within normal limits. Hand X-ray showed kissing delta phalanges (**Fig. 4A**), while feet X-ray depicted broad first metatarsal and great

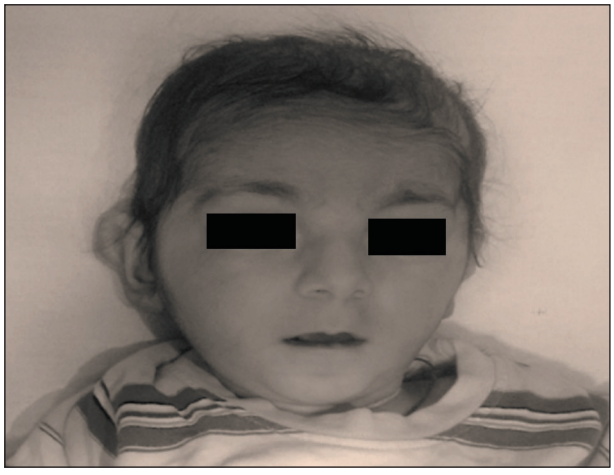

Figure 1. Microcephaly with distinctive facial features.

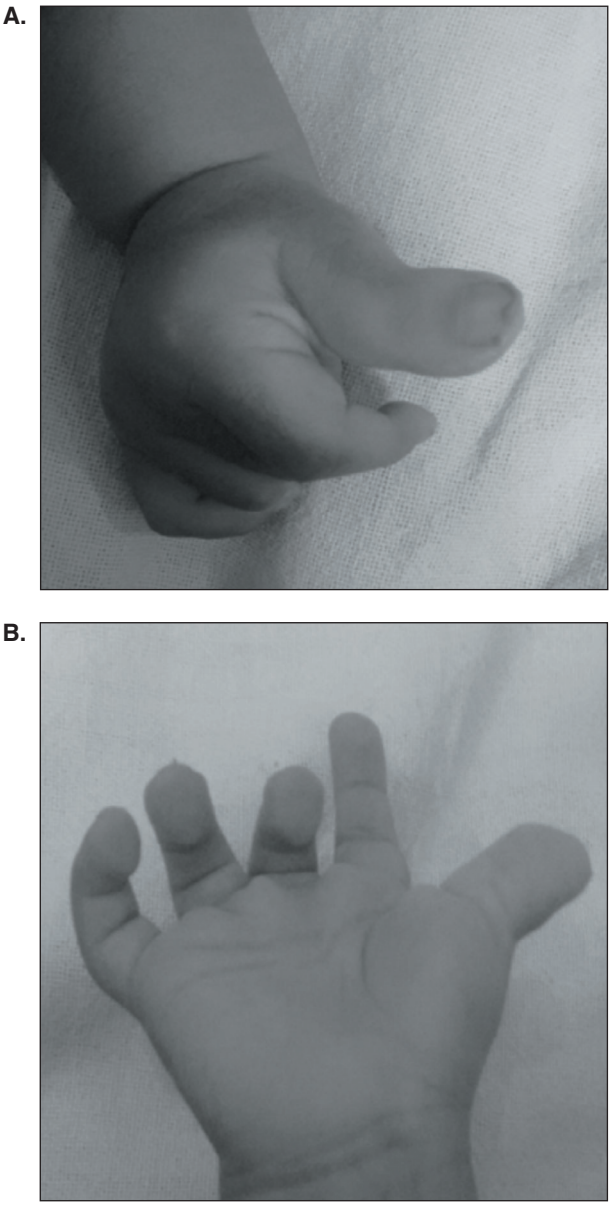

Figure 2. A. Broad thumb with radial angulation. **B.** Camptodactyly of 3rd, 4th and 5th fingers with persistent finger pads.

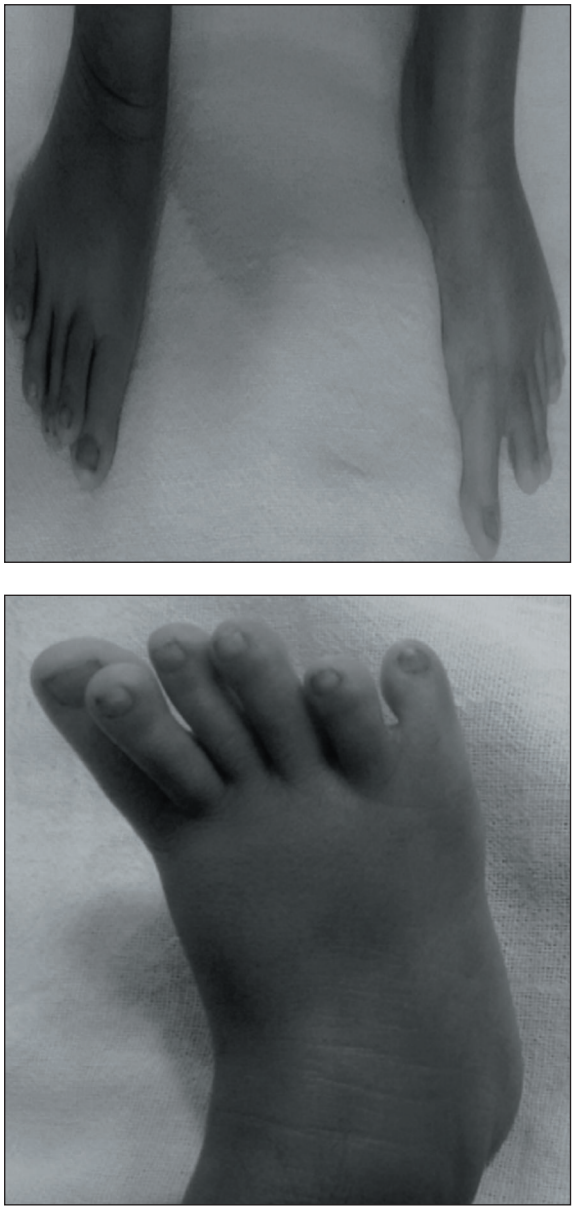

Figure 3. Bilateral postaxial polydactyly.

toe phalanges (**Fig. 4B**). Ultrasonography and MRI of abdomen (**Fig. 5**) was suggestive of horse shoe kidney with intra-abdominal testes.

Discussion

Our case was suspected to have RSTS based on characteristic facial features along with hand and feet anomalies. RSTS, also known as "broad thumbs syndrome" or "broad thumb-hallux syndrome", is characterized by the triad of broad and often angulated thumbs or halluces, distinctive facial features and intellectual disability. RSTS is inherited in an autosomal dominant manner. Mutations in *CREBBP* and *EP300* genes are known

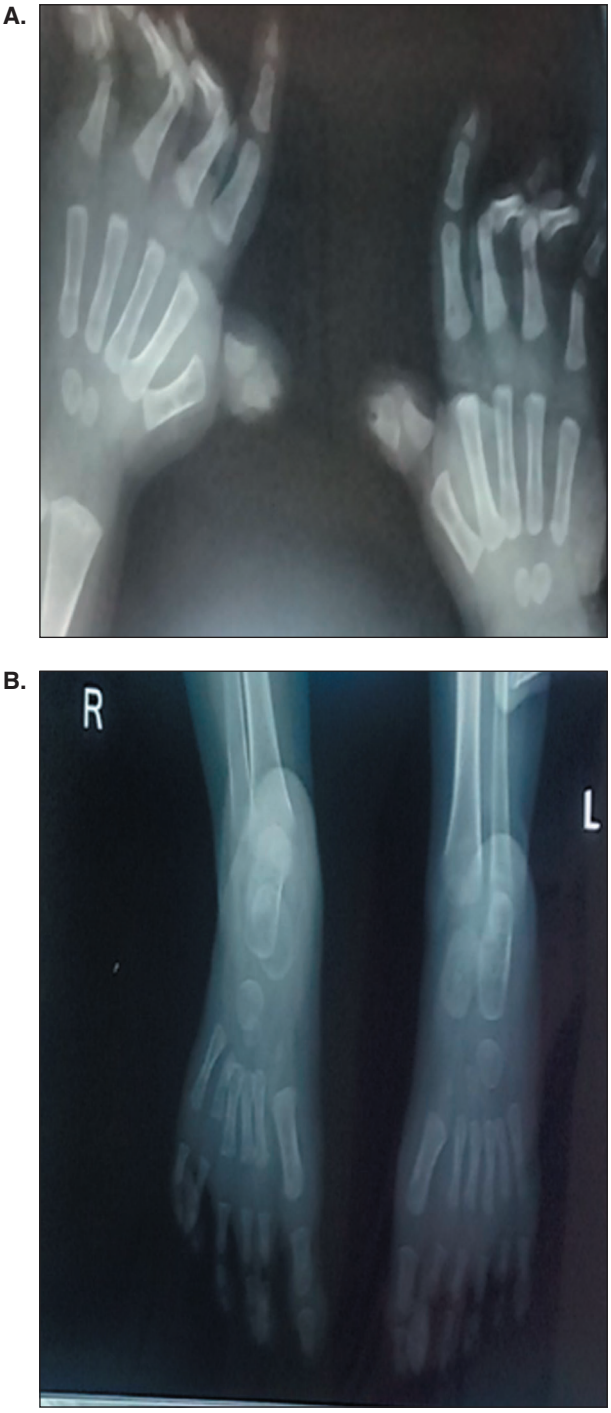

Figure 4. A. Hand X-ray showing kissing delta phalanges. **B.** Feet X-ray showing broad first metatarsal and great toe phalanges.

to be associated with RSTS. Most individuals with RSTS are sporadic cases, having unaffected family members or parents suggesting *de novo* mutation in affected individual, or as a result of germline mosaicism in one of the parents. Our case was also a sporadic case. Consistent with symptoms described by Cathy [4] and Rubinstein and Taybi [1], this child also shared similar craniofacial, skeletal and

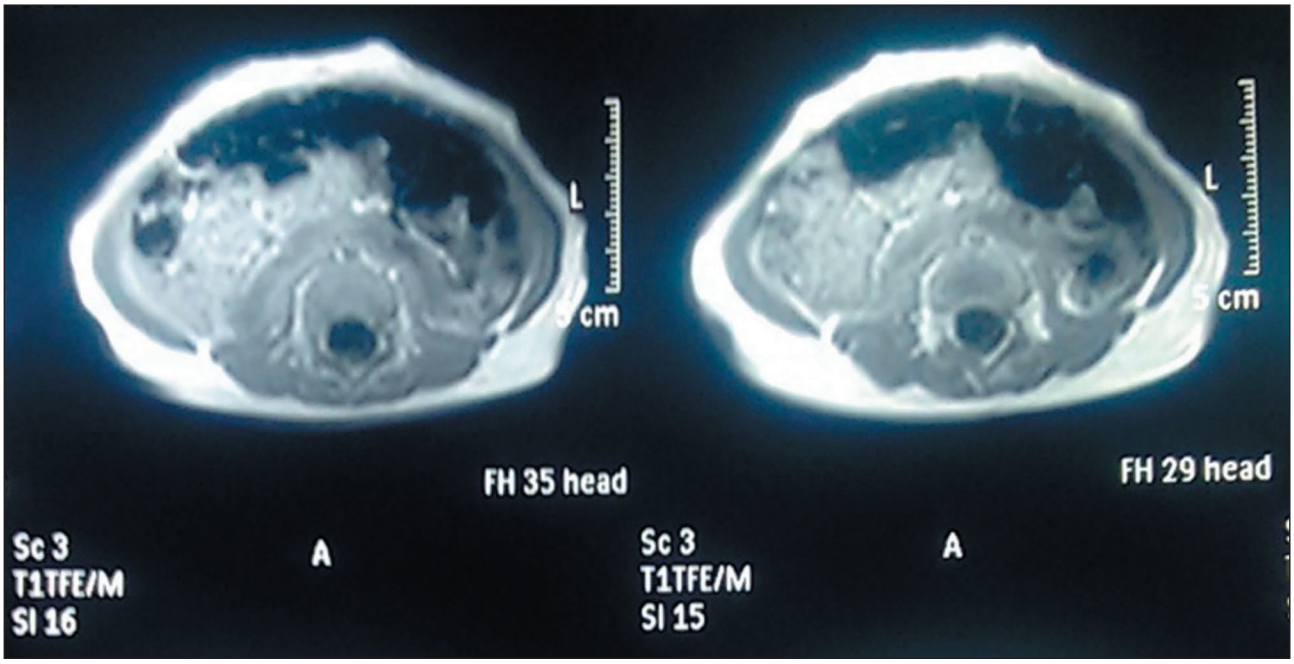

Figure 5. MRI of abdomen showing horse shoe kidney.

neurological features. The characteristics features of feet and hands are typically described as an enlarged 1st finger and clinodactyly of the 5th finger, whereas polydactyly has been rarely described in this syndrome [2]. Our case presented with bilateral postaxial polydactyly of feet. A constellation of symptoms including cardiovascular, dental, orthopaedic, eye, and genitourinary abnormalities have been described in this syndrome.

The diagnosis of RSTS is primarily based on clinical features, as these children have characteristic facial, hand and foot features. Diagnosis may be further supported by radiographic studies revealing characteristic malformations. Diagnosis can be confirmed by genetic testing, which involves sequence analysis of *CREBBP* gene or sequence analysis and/or deletion/duplication analysis of *EP300* gene [4]. In our case, genetic analysis was not performed as genetic testing is not currently available at our centre and parents could not afford the investigation owing to its high cost.

Management involves multidisciplinary approach, including surgical repair of digit anomalies, cryptorchidism, and supportive treatment for constipation. Child is under further evaluation for surgical management of digit anomalies. Early recognition of less common features of RSTS can help in its early diagnosis and thus in subjecting the patient to early intervention programme.

Conclusion

High degree of suspicion in a child presenting with multiple congenital anomalies has a great importance in diagnosis of a genetic syndrome like RSTS. Early detection is essential for prevention of morbidity, mortality and disability. This syndrome has various systemic abnormalities; a detailed and thorough clinical examination is essential in every child suspicious of RSTS syndrome. Whenever possible, genetic testing should be offered to every case, as it is needed to confirm diagnosis and is also important for prenatal testing for at risk pregnancy.

Declaration of interest

The Authors have no conflict of interest to disclose with regard to this article.

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