Case Report

Rubinstein-Tayabi Syndrome: A Rare Case Report

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Abstract

Rubinstein-Taybi syndrome (RSTS) is an extremely rare autosomal dominant genetic disease, with an estimated prevalence of one case per 125,000 live births. RSTS is characterized by typical facial features, microcephaly, broad thumbs and first toes, intellectual disability, and postnatal growth retardation. However, no standard diagnostic criteria are available for RSTS. In this review, we summarized the clinical features and genetic basis of RSTS and highlighted areas for future studies on an appropriate diagnostic protocol and follow-up care for RSTS.

Keywords: Broad Thumb-Hallux syndrome, Hypodontia, Rubinstein-Taybi syndrome.

Introduction

Rubinstein-Taybi Syndrome (RTS: OMIM 180849), or Broad Thumb-Hallux syndrome, was initially described by Michail *et al*. in 1957. In 1963, Rubenstein and Taybi reported on seven cases of this syndrome. Rubinstein-Taybi syndrome (RTS) is a rare developmental disorder characterized by craniofacial dysmorphisms, broad thumbs, and toes as well as mental and statural deficiencies.[1,2] RTS has a prevalence of 1 in 330,000 births and usually occurs sporadically, although it can be inherited as an autosomal dominant disorder. The diagnosis is based on characteristic features. Primarily characterized by poor post-natal height weight growth, intellectual disability, microcephaly, dysmorphic facial features, broad thumbs, and big great toes. Oral manifestations of this syndrome include limited mouth opening, a pouting lower lip, retro/micrognathia, a high arched and narrow palate, cleft uvula and palate, and rarely a cleft upper lip. Dental abnormalities occur in 67% of individuals with RTS and can include hypodontia, talon cusps, and enamel hypoplasia. Dental treatment is generally complicated due to difficulties in managing the patient's behaviour. In most patients with this syndrome, it is necessary to carry out the dental treatment under sedation or general anaesthesia.

Case Report

A 14-year-old girl accompanied by parents reported to the Department of Oral Medicine and Radiology, Seema Dental College and Hospital, Rishikesh, the UK with a complaint of yellowish discoloration of teeth. The following clinical extraoral findings were observed: stepping gait, short stature, broad thumb with malformed toes, hypermobility of arms, protruded chest, i.e. pigeon's chest (Figures 1-3). Intra-oral findings showed normal dentition. Mild horizontal opaque bands are seen on upper anterior suggestive of enamel hypoplasia. Calculus and plaque deposition could be noted on all teeth. After the complete clinical examination patient was referred for oral prophylaxis.

Discussion

Rubinstein-Taybi Syndrome is characterized by slow development of height and weight, microcephaly, dysmorphic facial features, broad thumbs, and great toes. The prenatal development is normal, with average or near-normal growth parameters at birth. The growth charts typically approach the lower limits of normality in the first postnatal period, primarily reflecting hypo-feeding exacerbated by gastroesophageal reflux. Subsequently, the tendency of overweight or obesity

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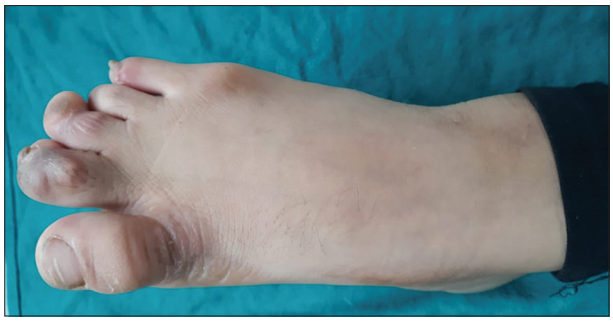

Figure 1: Broad thumb and malformed toes

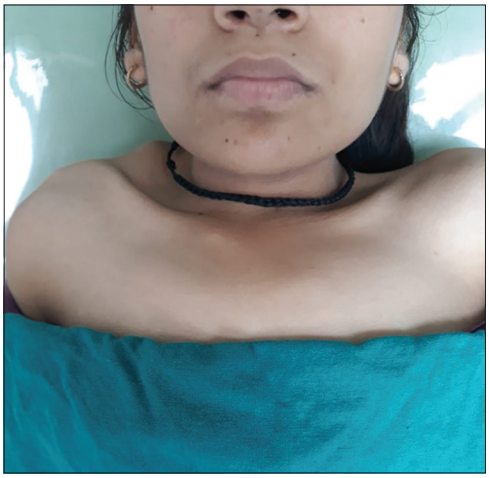

Figure 2: Pigeon chest

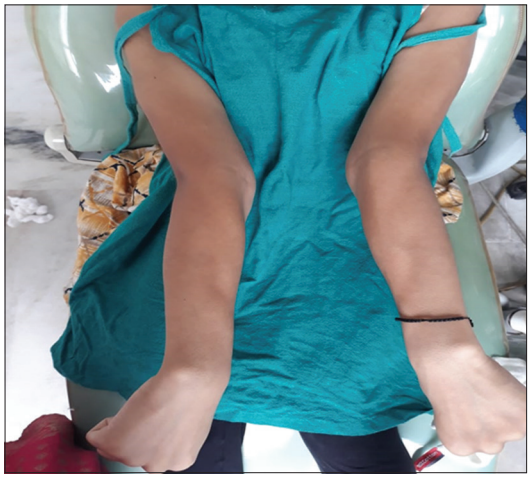

Figure 3: Hypermobility of arms

(earlier in males than females) can be observed during adolescence. Specific and recently reviewed growth charts are essential for appropriate assessment of the growth of affected individuals.[3,4] Facial features are primarily characterized by low frontal hairline, arched/thick eyebrows, downslanting of palpebral fissures, a protruding beaked nose with columella below alae nasi, dysplastic and low-set ears, an arched palate, mild micrognathia, dental anomalies (altered conformation, malocclusion, and overcrowding of teeth), and atypical smile ("grimacing"). The feet and hands typically present an enlarged first finger and clinodactyly of the fifth finger, whereas polydactyly with bifid thumbs and first toes is rarely observed. Other skeletal anomalies include abducted thumbs, vertebral anomalies, ligamentous laxity, severe and prolonged aseptic inflammation of the femur head, anomalies similar with Perthes disease (3%), and occasionally slipped capital femoral epiphysis. Particularly, high risk of cervical vertebral abnormalities (instability of C1–C2, os odontoideum, hypoplasia of the dens, fusion of the cervical vertebrae) has been reported with possible stenosis at the craniovertebral junction, which may cause cervical myelopathy.[5]

In most patients with this syndrome, it is necessary to carry out the dental treatment under sedation or general anesthesia. It is important to know that these patients might have upper respiratory obstruction during sleeping or sedation because of the anatomical characteristics of their maxillofacial region the treatment of the patients is performed under general anesthesia.[6,7]

Freitas *et al*. reported the periodontal and immunological status of a 14-year-old female patient with RSTS and possible association between syndrome and periodontal disease was reported in the paper. In this article, heavy calculus and plaque deposition could be noted on all teeth. The marginal gingiva was severely swollen and probing resulted in profuse gingival bleeding.[8,9]

It has been shown that mutations in the genes encoding the cyclic-AMP-regulated enhancer-binding protein (CREBBP) and the E1A-binding protein p300 (EP300) contributed to the development of RSTS. Therefore, genetic tests are useful for the diagnosis of RSTS, although most RSTS cases are currently diagnosed based on clinical features.[10,11]

Conclusions

RSTS is an extremely rare condition for which some clinical aspects have been clearly identified, but a lot of studies are ongoing and needed. Multicenter studies are needed to expand our knowledge on the clinical phenotype, identify specific genotype-phenotype correlations, evaluate the presence of somatic mosaicisms to better define mild phenotypes, and identify new candidate genes. The ultimate goal of these studies is to extend our current knowledge concerning this syndrome and to define new international guidelines for diagnosis, care and treatment of patients with RSTS.

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