From Diagnosis to Management: Rubinstein–Taybi Syndrome

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Rubinstein–Taybi syndrome (RSTS) or Broad Thumb-Hallux syndrome is a genetic disorder characterized by facial dysmorphism, growth retardation, and mental deficiency. Presented here is a review and a case report of the patient with RSTS with comprehensive oro-dental treatment. RSTS is a rare multiple congenital anomaly syndrome with only 600 cases reported worldwide. This patient presented typical features of RSTS which is a rare multiple congenital anomaly syndrome.

Keywords: Broad thumb, Expansion appliance, Rubinstein–Taybi syndrome, Talon’s cusp

INTRODUCTION

Rubinstein–Taybi syndrome (RSTS) or Broad Thumb-Hallux syndrome is a genetic multisystem disorder which is characterized by facial dysmorphism, broad thumbs and halluces, growth retardation, and mental deficiency. It was first described by Michael in 1957 in the French literature, and it is named after Jack Herbert Rubinstein and Rubinstein and Taybi. Both of them identified the constellations of a recognizable syndrome in 1963.

The incidence of the syndrome has been estimated to be 1 in every 3,00,000 newborns. A retrospective study conducted in genetic and metabolic unit of a tertiary care teaching hospital in North India over a period of 3½ years reported 11 cases of RSTS. There is an equal male and female incidence. The cause of the syndrome remains unknown. The genetic profiling of this syndrome states that CREB binding protein (CREBBP) and EP300 are the only genes currently known to be associated with RSTS. It results due to de novo mutation, and in most cases, the parents of an individual with RSTS are unaffected.

Mental deficiency is characteristic, and there is severe delay in expressive speech. Average IQ reported in these cases is 51. Electroencephalographic abnormalities, seizures, the absence of corpus callosum, and hyperactive deep tendon reflexes have been reported. Incomplete or delayed descent of the testes has also been seen in males.

The other characteristics 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 of the upper lip. Dental abnormalities occur in 67% of individuals with RSTS and can include hypodontia, retained primary teeth, talon cusps, and enamel hypoplasia. An increased rate of caries and periodontal disease has been reported in these patients although periodontal disease has not always been associated with this syndrome (Table 1).

Presented here is a case report of the patient with RSTS with comprehensive oro-dental treatment.
A 10-year-old boy accompanied by his parents reported to the Department of Paedodontics and Preventive Dentistry, Subharti Dental College, Swami Vivekanand Subharti University, Meerut, with a chief complaint of irregular teeth. The diagnosis of RSTS was made following a complete clinical examination including X-rays of the hands, a computed tomography-scan, and an electroencephalography to evaluate the electrical activity of the brain. It was furthermore confirmed by genetic testing for a deletion or mutation in chromosome 16p and by the presence of characteristic features. The prenatal and natal history of the child was uneventful. He was the first child of a non-consanguineous couple. His younger brother is normal and has no sign of this syndrome and of any dental anomaly. The clinical history of the patient revealed that there was delay in developing milestones. The child was extremely cheerful and cooperative on interaction and did not have any problem in socializing and attending school, presently studying in the fourth standard. The patient appeared mentally challenged and hyperactive. IQ evaluation was done using sequin form board test which revealed the patient to be borderline.

The patient had a distinctive facial appearance with a broad forehead, hypertelorism, broad nasal bridge, beaked nose, thin upper lip, and pouting lower lip (Figure 1). The patient had short stature (Figure 1) with broad thumbs (Figure 2) and dry skin. No associated cardiac abnormalities were found.

Intra-oral findings revealed crowding of teeth (Figure 3), a high-arched V-shaped narrow palate (Figure 4). On examinations, the presence of talon cusps was seen on both maxillary central incisors (Figure 4). The patient had no caries although there was deposition of heavy calculus and plaque on all teeth, especially on the lingual surface of lower anteriors.

After the complete clinical examination and investigation, dental treatment plan was derived. The entire treatment was planned in the dental operatory without the use of general anesthesia owing to the patient’s cooperative behavior, and entire treatment was done in multiple sitting
in the dental operatory. The first visit included establishing the rapport with the child, oral prophylaxis, and fluoride application. The second visit constituted impression making with alginate. In second visit alginate impression was made. Therefore, a maxillary impression was taken after using local anesthetic-flavored jelly on the palate, and extraction of 83 was also done on the same day. The third visit constituted the delivery of the expansion appliance for correction of V-shaped arch (Figure 5) and grinding of talon’s cusp followed by Fluoride application. Detailed oral hygiene instructions were given to his mother who provided routine care for the patient, and the patient recalled follow-up sessions every 3 months.

DISCUSSION

RSTS is a rare multiple congenital anomaly syndrome with only 600 cases reported worldwide. This patient presented typical features of RSTS which is a rare multiple congenital anomaly syndrome. Other syndromic entities that may give confusion are Saethre–Chotzen syndrome, Cornelia de Lange syndrome, and trisomy 13. The facial features also show resemblance to Floating-Harbor syndrome and Gorlin-Chaudhry-Moss syndrome. Extraoral features of this patient were a broad forehead, hypertelorism, broad nasal bridge, beaked nose, and thin upper lip. There was the presence of dry skin. Intraoral features included crowding of teeth, a high-arched V-shaped narrow palate, pouting lower lip, the presence of talon cusps on both maxillary central incisors with no caries but deposition of heavy calculus and plaque, especially on the lingual surface of lower anteriors. It was reported that patients with this syndrome have an increased rate of caries (15-36%) because of their poor oral hygiene, which was not similar to our patient as the mother was vigilant about the child’s oral health and adequate oral hygiene methods were adopted. Malposition and crowded teeth are present in 62% of patients which is similar to this patient. Malpositioning of teeth was one of the important problems in this patient. Hennekam and Van Doorne also suggested that hypodontia, malformed teeth, natal teeth, and hyperdontia can be manifested in this syndrome. This patient had no supernumerary teeth. There was the presence of talon’s cusp in both upper central incisors. Gardner and Girgis have observed talon cusps in over 90% cases whereas Hennekam and Van Doorne after studying 45 cases of RSTS in Netherlands reported that 92% of them had talon cusps. Talon cusps are also present in 1% of normal population.

Treating children with RSTS is a challenge. In most patients, it is necessary to carry out the dental treatment under sedation or general anesthesia. These patients may have complications associated with a certain muscle relaxant (succinylcholine) and certain anesthesia. These children may have higher rate of cardiac, physical, and conduction abnormalities which may cause unexpected results with cardioactive substances. 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. This patient was extremely cooperative; therefore, his treatment was done in the dental operatory without the use of any of these advanced behavior guidance methods. The rarity of the RSTS coupled with the fact that this case presents with dry skin which was confirmed after consultation from Department of Dermatology in Subharti Medical College and child’s exceptionally cooperative level (Frankel ++ behavior). The presence of these two features makes this case an interesting one.

Petru et al. suggested that RSTS results from chromosomal rearrangement of chromosomes 16p but also from point mutation in CREBBP gene.
CREBBP and EP300 are the only genes associated with syndrome.\textsuperscript{20,21} The locus of the gene for RSTS may be situated on 16p13.3.\textsuperscript{22}

In our patient, genetic testing report revealed that there was deletion in chromosome 16p which confirmed that the patient was suffering from RSTS. In most patients with such syndromes, the emphasis on care required is on the primary disabling condition, and so, they often receive inadequate dental care. This is particularly unfortunate since oral health and function are essential to the overall health and well-being of growing children, including those with disabilities. The comprehensive care required by these patients is time-consuming, and many patients as well as their families are uncooperative. Therefore, the responsibility of the dentist lies beyond addressing the only chief complaint of these special children.

CONCLUSION

We found that no other studies describing RSTS patients with borderline IQ, patient cooperation, and dry skin in literature. In our case, the patient IQ was borderline. Hence, more number of cases should be analyzed for IQ level and dental treatment protocol.

REFERENCES


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