CASE REPORT

Rubinstein–Taybi syndrome: A pediatric case report

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Introduction
Michail et al. in 1957 initially described Rubinstein–Taybi syndrome (RTS).\(^1\) In 1963, Rubenstein and Taybi reported on approximately seven cases, which included a group of congenital anomalies consisting of short, broad thumbs and great toes, mental retardation, high-arched palates, particular facial abnormalities, and histories of recurrent respiratory infections.\(^1\)

Incidence of the syndrome has been estimated to be in 1/125,000–300,000 individuals.\(^2\) Male and female shows equal incidence.\(^3\-^4\) RTS shows autosomal dominant mode of inheritance which involves mutations of genes encoding cAMP-regulated enhancer binding protein (CREBBP) and E1A-binding protein p300 (EP300). Therefore, for the diagnosis of RTS, genetic tests are useful.\(^5\)

Case Report
A girl aged 9-year-old reported to the department of pedodontics and preventive dentistry with a complaint of dental caries and bleeding of gingiva. The patient had delayed physical and mental development. There was no significant family history. The patient was diagnosed with Rubinstein–Taybi syndrome at the age of 5 years.

Extraoral findings of patients were slanting palpebral fissures, hypertelorism, beaked shaped nose, deviated nasal septum, and small size mouth [Figure 1]. Intraoral findings were high-arched palate, significantly small mouth opening, collapsed bite, and malocclusion [Figure 2a and b]. Broad thumbs, broad terminal phalanges were absent [Figure 3]. The patient had multiple grossly carious lesions and gingivitis with profuse bleeding on probing [Figures 2a and b, 4]. Orthopantomogram of patient showed oligodontia with missing 31, 32, 33, 41, 42, 43, 12, and 22 [Figure 5].

Preventive dental health strategies with parental counseling were initiated in this patient. Extraction of all grossly decayed teeth followed by restorations and extensive oral health care was planned for the patient. While treatment, behavior modification techniques were used such as tell-show-do, physical restraints with the positive reinforcement, and involvement of mother.

Discussion
Rubinstein–Taybi syndrome is a rare multiple congenital anomaly caused by either a microdeletion at 16p 13.3 or mutations in the CREBBP or CBP or EP300 gene (at 22q13).\(^6\) Although some mutations found in CREBBP are translocations, inversions, and large deletions, most are point mutations or small deletions and insertions. These two genes encode histone acetyltransferases (HAT’s), which are transcriptional coactivators involved in many signaling pathways. Loss of HAT activity accounts for the phenomena seen in Rubinstein–Taybi patients.\(^7\-^11\) Dentists along with the medical team need to differentiate this syndrome
Rubinstein–Taybi Syndrome

with other conditions such as Down’s syndrome, Floating-Harbor syndrome, and Pfeiffer and Saethre-Chotzen syndromes. Clinically, this syndrome is characterized by decreased growth, mental retardation, and broad mediated deviated thumbs with big toes. Patients also exhibit some craniofacial abnormalities such as downward slanted palpebral fissures, microcephaly, posterior rotated ears, hypertelorism, long eyelashes, pouting upper lip, and beaked nose with columella protruding below alae nase. Other clinical features include cryptorchidism, gastrointestinal tract abnormalities, and recurrent respiratory tract infections. Congenital anomalies of cardiovascular system are also described such as arterial septal defects, patent ductus arteriosus, ventricular septal defect, coarctation and stenosis of aorta, and pulmonic stenosis. She did not present any cardiac, gastrointestinal tract, or respiratory abnormalities, which are frequently found in 24–38% of RTS children.

Various studies have shown that 67% of individuals with RTS present with dental abnormalities. These anomalies include restricted mouth opening, retro/micrognathia, pouting of lower lip, high arched and narrow palate, cleft palate, and rarely cleft lip. Hypodontia or oligodontia, retained primary teeth, talon cusps, and enamel hypoplasia are common occurrence in RTS. An increased rate of caries and periodontal disease has been reported in these patients (15–36%) because of their poor oral hygiene and inability due to mental retardation which was similar to our patient. In this patient, reduced mouth opening, oligodontia, and high-arched palate were present. Generalized heavy calculus and plaque deposition was present due to very poor oral hygiene.

Figure 1: Facial characteristics include hypertelorism, slanting palpebral fissures, and beaked nose with deviated nasal septum

Figure 2: (a) Maxillary arch shows high-arched palate with multiple grossly carious lesions, (b) Small mouth opening showing malocclusion with oligodontia

Figure 3: Broad thumbs, broad terminal phalanges were absent

Figure 4: Lower molar teeth also had several grossly carious lesions and lower anteriors were missing

Figure 5: Orthopantomogram showing oligodontia with missing 31, 32, 33, 41, 42, 43, 12, and 22
with generalized gingivitis and localized periodontitis. Upper and lower primary teeth had grossly carious lesions.

Mental development is affected in RTS patients, so the level of cooperation is less among them compared to the normal ones. Many patients with RTS may require dental treatment to be carried out under sedation or general anesthesia, depending on their level of cooperation.\(^\text{[17]}\) Anesthesia has to be considered under strict medical supervision in a hospital setup. The treatment in RTS patients demands palliative care and preventive management strategies. This should be followed by various other oral rehabilitation treatment procedures such as restorations, extractions, preventive, and interceptive orthodontic management. Genetic counseling may be considered as a part of counseling to parents.

**Conclusion**

The orodental complications of RTS are important but usually remain neglected aspects of the disorder. It has become apparent that the multifaceted orodental problems necessitate a team approach toward dental management which may require interventions under anesthesia. Multidisciplinary collaboration is crucial for effective management and for the well-being of the affected person.

**Clinical Significance**

We present this case report to inform the pediatric community of this condition and document a rare syndrome. These types of case reports add more literature in the path of diagnosing and increasing the knowledge of various clinical features and prevalence of this syndrome.

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