CASE REPORT

Rubinstein–Taybi syndrome: A case report

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Abstract
Rubinstein–Taybi syndrome (RTS) is characterized by growth moderate-to-severe intellectual disability, typical face, and particular craniofacial characteristics along with broad thumbs and first toes. The prevalence at birth was estimated to be 1 in 125,000 living newborn infants. The purpose of this case report was to demonstrate the oral and dental manifestations of a 6-year-old girl suffering from RTS.

Keywords: Prognathic maxilla, retrognathic mandible, Rubinstein–Taybi syndrome

Introduction
Originally described in 1963, the Rubinstein–Taybi syndrome (RTS) is a well-defined entity characterized by growth moderate-to-severe intellectual disability, typical face, and particular craniofacial characteristics along with broad thumbs and first toes.¹ The prevalence at birth was estimated to be 1 in 125,000 living newborn infants. Linked to chromosome region 16p13.3 by coincident structural chromosomal disruption of that band RTS has been shown to be associated with microdeletions at band 16p13.3 in 25% of cases studied by fluorescence in situ hybridization (FISH) with cosmid probe RT1.² RTS presents at birth, with certain characteristics becoming more apparent as the patient ages. Prenatal growth is normal, but growth retardation is apparent within a few months of birth and requires attention to avoid overfeeding.³ The purpose of this case report was to demonstrate the oral and dental manifestations of a 6-year-old girl suffering from RTS.

Case Report
A 6-year-old female reported to the Department of Orthodontics and Dentofacial Orthopaedics of a tertiary care government institute with the chief complaint of increased upper gums and difficulty in closing mouth.

A case history revealed a C-section birth with low birth weight (2 kg), the patient was admitted in neonatal intensive care unit for 5 days after birth. The patient was also unable to breastfeed and later mental retardation was also detected with age. She was also unable to hear for which she was operated for cochlear implant at the age of 4 years. A history of genetic evaluation has confirmed microdeletion in 16p13.3.

Clinical examination of the patient displayed retarded growth with height 88 cm and weight 13 kg. The patient was able to hear and responsive to name but was not able to fully understand and follow instructions.

On extraoral examination, it was found that the patient had a leptoprosopic face, hypertelorism, broad nose, absent philtrum, and increased gingival shows at rest with incompetent lips [Figure 1a]. She has a convex profile, acute nasolabial angle, and clinical appearance of vertical maxillary excess with retrognathic mandible and both lips placed anterior to E-line [Figure 1b].

On intraoral examination, it was found that she was in early mixed dentition with all permanent first molars and lower right central incisor erupted, narrow maxillary arch, and a high-arched palate. Flush terminal deciduous molar relation and end on permanent molar relation bilaterally with increased overjet as measured from canines and 100% deep bite with apparent vertical growth pattern [Figure 1c-e].

Radiographic examination confirmed a marked convex profile, short cranial base, prognathic maxilla, retrognathic mandible, and abnormally curved lower border of corpus of mandible with vertical growth pattern [Figure 2a-c].
However, this patient did not exhibit broad and angulated thumbs and hallucs [Figure 3a-d].

Discussion

A breakthrough in the search for a specific cause in RTS came with the identification of cytogenetic rearrangements involving 16p13.3 in few patients with classical manifestations of RTS. McGaughran et al. found two deletions in a group of 16 patients from the United Kingdom and Wallerstein et al. showed seven deletions out of North American cohort of 64 RTS patients.

In 65% of the cases, RTS is caused by cAMP response element-binding protein mutations on 16p13.3, in 3–5% of the cases by EP300 (E1A-binding protein, 300-kDa) mutations on 22q13 and in the remainder of the cases, its cause remains unknown.

RTS patients typically have a normal prenatal course and are recognized at birth due to characteristic face, hand, and feet anomalies. Mental retardation in RTS ranges from mild to severe, the mean intelligence quotient (IQ) being around 35. Over 50% of patients have an abnormal Electroencephalogram (EEG), with roughly 28% of them developing seizures. A third of RST patients have congenital heart defects. The features and characteristics of RTS as described in Online Mendelian Inheritance in Man are as follows:

- Autosomal-dominant inheritance
- Short stature with average adult male height 153 cm and average adult female height 147 cm. Postnatal growth retardation but usually develops obesity after puberty
- Microcephaly, large anterior fontanel, late closure of fontanel, and frontal bossing. Low anterior hairline, hypoplastic maxilla, micrognathia-retrognathia, grimacing or unusual smile with almost closing of the eyes are some other characteristics. Low-set ears, hearing loss, and recurrent otitis. Heavy eyebrows, high-arched eyebrows, long eyelashes, ptosis, epicanthal folds, strabismus, nasolacrimal duct obstruction, cataracts, glaucoma, coloboma, down slanting palpebral fissures. Beaked nose, deviated nasal septum, and broad nasal bridge. Small opening of the mouth, narrow palate, and high-arched palate. Dental crowding, talon cusps, crossbite, screwdriver permanent incisors, enamel hypoplasia, and enamel discoloration
- Recurrent respiratory infections and sternal anomalies. Delayed skeletal maturation and scoliosis. Broad thumbs with radial angulation, fifth finger clinodactyly, persistent fetal fingertip pads, syndactyly, polydactyly, and single transverse palmar creases
- Mental retardation (average IQ 51), agenesis of corpus callosum, severe expressive speech delay, poor coordination, EEG abnormalities, seizures, and hypotonia. Good social contacts, short attention span. About 10% of cases are secondary to submicroscopic deletions of 16p13.3 detectable by FISH. A small minority of patients have translocations and inversions involving 16p13.3. There is de novo mutation in most cases with variable severity.
Dental management

Oral hygiene technique recommendations were made to both the parents at follow-up sessions for every 6 months. The patient was consulted for orthodontic treatment. Optimal patient cooperation was lacking. Hence, initially, a maxillary slow expansion appliance with a Schwartz appliance has been prescribed to the patient. The patient is on regular follow-up.

Conclusion

The maxillofacial and dental features of RTS are important but neglected aspects of the disorder. Long term follow up is essential for effective management and for the well-being of individuals affected with RTS. This case report endeavours to add more literature in the knowledge of various clinical features and management of this syndrome.

References