

Rubinstein-Taybi syndrome: case report

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Abstract

A patient with Rubinstein-Taybi syndrome is presented with the characteristic clinical features including small and short stature, severe mental retardation, and small maxilla with a dental malocclusion. Treatment for this patient was accomplished utilizing a general anesthetic. These special patients who lack the ability to accept dental treatment in the normal situation require extensive and comprehensive dental treatment compared with nonhandicapped patients.

Rubinstein and Taybi first described the clinical features of Rubinstein-Taybi syndrome (RTS) in 1963. The diagnostic clinical features present at birth include broad thumbs and toes, distinct facies, and mental retardation. The facial appearance of the newborn has the following characteristics: beaked nose and broad nasal bridge with the nasal septum extending below the alae, epicanthal folds, microcephaly, palpebral fissures which slant downward, prominent forehead and strabismus (Fig 1). Grimacing or an unusual smile may be observed (Goodman and Gorlin 1983). Skeletal abnormalities observed are small and short stature, retarded osseous maturation, hypoplastic maxilla with narrow



FIG 1. Facial features include slanting palpebral fissures and an unusual grimacing smile.

palate and dental malocclusions, auricles low set and/or malformed, and cryptorchidism (Wiedeman et al. 1985).

In the neonate this syndrome can be confused with the de Lange syndrome, trisomy 13, Apert syndrome, and Pfeiffer syndrome. Therefore, it is best to consider

individuals without all the classic features of RTS syndrome as having an "incomplete form" rather than an incorrect diagnosis (Goodman and Gorlin 1983). Feeding problems, upper respiratory infections, and recurrent otitis media are common during infancy.

A dental anomaly frequently seen in RTS patients is talon cusps (Mellor and Ripa 1970). A talon cusp is best defined as an abnormally enlarged cingulum on a maxillary incisor tooth (Gardner and Girgis 1979). Finding talon cusps in the normal population is rare—its occurrence in the RTS patient appears clinically significant.

The frequency of RTS occurrence in the mentally retarded institutionalized population older than 5 years of age has been estimated at 1/300-500 (McKusick 1886). The frequency of this syndrome occurring in the population at large is unknown (Padfield et al. 1968). All RTS patients exhibit degrees of language, mental, motor, and social retardation with the most usual IQ being in the 40-50 range (Smith 1982). Although the etiology of this syndrome is unknown, many affected patients tend to resemble other but unrelated children who are victims of the same syndrome (Baker 1987).

Case Report

An 8-year-old white male with RTS, the youngest of a 2-child family, was referred to the dental clinic at Children's Mercy Hospital in Kansas City, Missouri, for a dental evaluation.

The following clinical findings, consistent with an RTS diagnosis were observed: (1) small and short stature with broad thumbs (Fig 2); (2) severe mental retardation; (3) a small maxilla with numerous carious lesions (Fig 3); (4) severe crowding with a bilateral posterior cross-bite (Fig 4); and (5) anterior cross-bite with no evidence of talon cusps (Fig 5).

The patient's weight was at the 20th percentile and his height below the 5th percentile for boys 2-18 years of

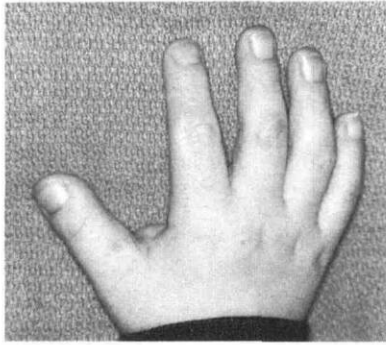


FIG 2. Broad thumbs are characteristic of the syndrome.

age (Hamill et al. 1979).

In consideration of the patient's ability for cooperation and acute situational anxiety, all necessary dental treatment was performed under general anesthesia.

Treatment

The patient was admitted to the Same Day Surgery Clinic at Children's Mercy Hospital for dental rehabilitation. The physical examination was within normal limits for a patient with RTS and revealed no cardiac abnormalities. Therefore, reports of cardiac arrhythmias produced by succinylcholine in some patients with RTS was of minimal concern (Stirt 1982). After evaluation by anesthesia department staff, the patient was rated as a status II (American Society of Anesthesiologists) patient and an acceptable risk for general anesthesia.

The patient was taken to the operating room and was administered a general anesthesia. All the carious teeth were either extracted or restored with silver amalgam restorations or stainless steel crowns. The patient tolerated this procedure well and experienced an uneventful recovery.

Discussion

Most RTS patients are so severely retarded that they are confined in mental institutions. The present report of an 8-year-old white male with RTS who resides at home with an older normal sibling was presented to show the dental needs and requirements of these special patients. The dental treatment was accomplished in a single appointment utilizing general anesthesia.

A 3-month recall program for this patient was estab-



FIG 4. An anterior and bilateral posterior cross-bite was observed.

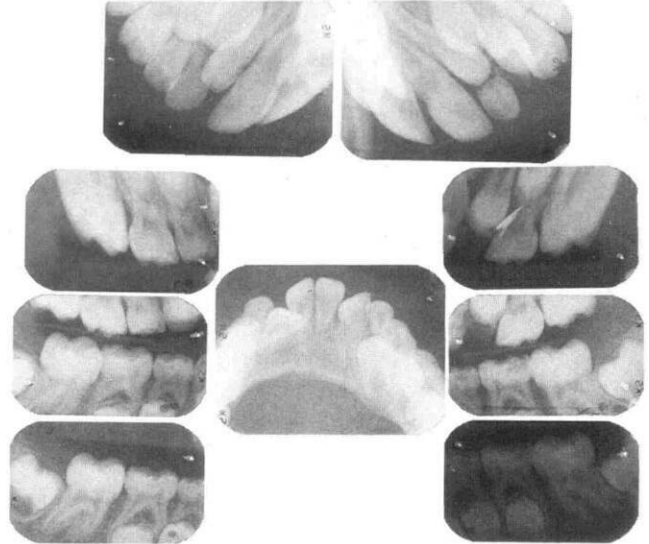


FIG 3. Radiographs demonstrating numerous carious lesions.

lished to monitor closely his dental care requirements and aid in intercepting and correcting any dental problems as they occur. With continuing parental support and supervision this patient will become capable of functioning to his maximum potential in the home environment.

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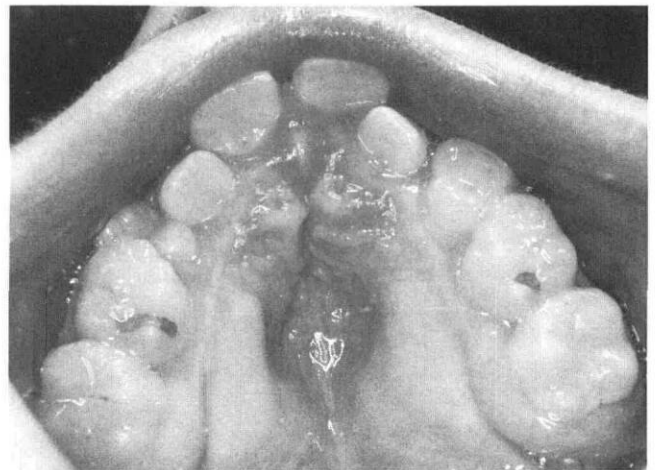


FIG 5. The maxillary incisors show no evidence of talon cusps.

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