# Respiratory arrest in Treacher-Collins syndrome: implications for dental management: case report

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Treacher-Collins syndrome (TCS) or mandibular dysostosis, is a rare autosomal dominant craniofacial malformation that has been described extensively in the scientific literature,[1] with more than 250 cases reported.[2] Most reports to date have focused on the facial morphology of this syndrome: antimongoloid slanting of palpebral fissures, malformed auricles, mandibular and zygomatic bone hypoplasia, receding chin, and others.[2] More recent studies[1,3-5] expressed a particular interest in the morphology of anatomical structures related to the upper airway in these patients that can develop early respiratory problems.[5] In numerous reports of TCS patients with a history of respiratory failure, the phenomenon had been attributed to glossoptosis, or to the "lethality of the gene", rather than to an actual restriction of the entire pharyngeal airway.[1] In their study using videofluoroscopy, Shprinzen et al.[1] disclosed marked narrowing of the airway in 11 patients with TCS. In several of them, the pharynx was less than 1 cm in width, and it was thought that the reduced airway in these patients might help to explain the frequent reports of neonatal death associated with the syndrome. The researchers concluded that pharyngeal hypoplasia is a primary feature of the TCS. Johnson et al.[3] discovered that patients with TCS are at high risk for upper airway obstruction with associated symptoms like sleep apnea due to the micrognathia that causes the tongue to fall posteriorly onto the oropharynx and obstruct the airway. In a recent comprehensive study, Arvystas and Shprinzen[5] gathered the applicable information concerning the life-threatening upper airway compromise in TCS and related it to the combination of basicranial kyphosis, narrowing of the pharynx, and the severe retrognathism and ramal height defficiency. Dental treatment on posterior teeth in patients with TCS may be extremely difficult since the mandibular defects dictate a very narrow mouth opening.

The purpose of this paper is to report two episodes of the dental treatment of a 7 1/2-year-old boy with TCS. One was respiratory arrest during ambulatory treatment, and the other, a difficult intubation at subsequent session under general anesthesia. The events and recommendations for treatment are discussed.

## Case report

A 7 1/2-year-old boy with TCS was scheduled for ambulatory dental treatment at the department of pediatric dentistry, Hadassah Faculty of Dental Medicine, for the first time. The four permanent molars needed restorations.

### Past medical history

The child had had four sessions of general anesthesia in the past 5 years: two sessions of surgical procedures for correcting defects in his face (bone grafts to the orbital floor, and correction of the antimongoloid obliquity of his eyes), and two sessions for dental treatment. In all the general anesthesia sessions, intubation was very difficult. The child was deaf and suffered from speech problems probably due to his hearing loss.

### Management

We originally planned to premedicate the child because of apprehensive behavior. The child arrived late, so the doctor decided to use nitrous oxide/oxygen only. Radiographs were to be exposed after sedation initiation. The child was afraid of the nasal mask due to unpleasant experiences with the several operations under general anesthesia; therefore, dental treatment started using conventional nonpharmacologic behavior management without nitrous oxide/oxygen analgesia. The treatment was planned to begin with the mandibular left first permanent molar, which required a stainless steel crown.

### Dental treatment

One carpule (1.8 cc) of 2% xylocaine with 1:100,000 epinephrine was administered for local anesthesia, and a rubber dam placed. While preparing the tooth, the dentist noticed that the child gradually stopped his mild crying and became completely quiet. The child's chest showed no observable breathing and his face was

cyanotic. The rubber dam was quickly removed, the child placed in the supine position, and manual ventillation using a nasal mask with 100% oxygen was performed. A second operator introduced an airway with maximal lifting of the chin anteriorly. These actions immediately resolved the situation, just before the arrival of the emergency team. Dental treatment was terminated. Because of the child's dental anxiety, past dental experience with its associated apprehensive behavior, and after consulting with the parents, dental treatment under general anesthesia was chosen. During the discussion with the parents about the continuation of treatment, they revealed that the child, when in disciplinary confrontation with his parents, had initiated several instances of breath holding, similar to that seen during the dental treatment, but not so severe.

### The general anesthesia

In the process of general anesthesia, despite the use of a fiber-optic airway instrument, intubation was extremely difficult due to the small mandible and its retroposition. The profile of the child can be seen in Fig 1. This type of profile is typical of TCS, and resembles

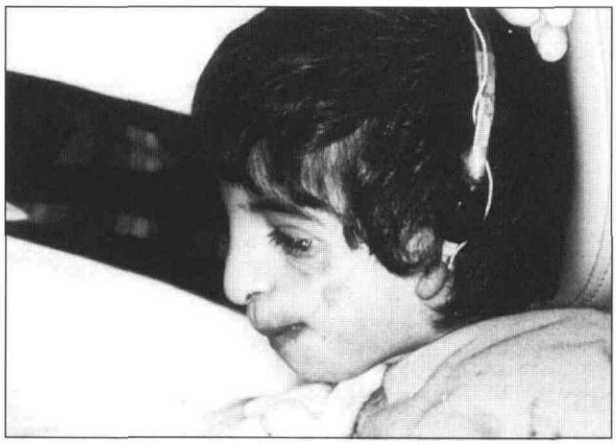

**Fig 1: Treacher-Collins syndrome in a 7 1/2-year-old boy. Note the depressed cheek, deformed auricle with a hearing aid, retrognathic hypoplastic mandible, fish-like appearance of the mouth, and anti-mongoloid slant of the eyelids. Also note the scalp hair onto the lateral cheek.**

previous reports.[3,5,6] A lateral radiograph of the head and neck demonstrates the retrognathic mandible (Fig 2). The treatment plan included restorations on teeth 3, 14, 30, A, and B, and the stainless steel crown on tooth 19. All these procedures were done with difficulty because of the narrow opening of the child's mouth, even under general anesthesia. Detailed oral hygiene instructions, and a thorough follow-up protocol were given to the parents.

## Discussion

An anxious and apprehensive child patient with TCS, whose previous dental treatments were done under general anesthesia, presented a dilemma to the dentist. On one hand, because of the previous exposure to general anesthesia and the definite need for future operations under general anesthesia to correct the syndrome-associated problems, minimizing the number of general anesthesia exposures was desirable. On the other hand, ambulatory treatment might have required some kind of sedation. The micrognathia and retrognathia associated with TCS are themselves major risk factors for airway blocking, and any premedication may enhance development of respiratory depression. In this case, we decided to try ambulatory treatment. Avoiding premedication turned out to be an advantage when the respiratory arrest occurred in that there was no possible pharmaceutical cause. The airway obstruction was detected by monitoring the chest movements and observing the color of the lips. A pulse oximeter might have been useful in this case. The pulse oximeter provides an early warning of desaturation long before the patient develops clinical cyanosis or changes in vital signs.[7] It should be noted that 5.0 g of desaturated hemoglobin are required to produce visible cyanosis, which converts to a saturation level below 70% in most patients—well before the point at which the clinician wishes to intervene.[8] A pulse oximeter, or at least a precordial stethoscope, can be a useful adjunct for TCS patients, to prevent obstructive airway complications. Jaw positioning and airway management are primary recommendations that ought to be

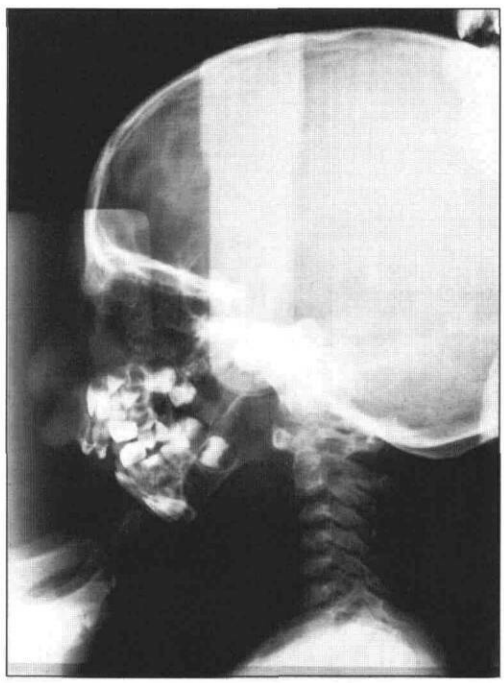

**Fig 2: A lateral radiograph of the boy with TCS at age 4 years, showing micrognathic retruded mandible with deficient ramus and obtuse angle, associated with open bite. The undersurface of the mandible is pronouncedly concave.**

made for routine care. The risk potential of sedation, which might compromise the protective reflexes, must be seriously considered.[9]

In Potsik's comment on an article dealing with airway obstruction and sleep apnea in children,[10] he calls attention to the fact that airway obstruction with severe obstructive apnea is not rare, and may occur from a variety of causes in patients with craniofacial malformations. He emphasizes that we are unaware of its true incidence and most specialists and primary care physicians fail to include historical data about breathing in their routine evaluations.

A more general conclusion of this report is the immense importance of prevention in young patients with TCS. As with any other special care patients, dental health education, and frequent preventive treatment must be encouraged to preclude having to treat the patient too often, and to avoid the dilemma of choosing the best of two compromised treatment modalities—both carrying potential risks to the patient with TCS.

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