# Obstructive Sleep Apnea in Treacher-Collins Syndrome

**CYNTHIA JOHNSTON, M.D.**
**LYNN M. TAUSSIG, M.D.**
**CHARLES KOOPMANN, M.D.**
**PHILIP SMITH, D.D.S.**
**JOHN BJELLAND, M.D.**
*Tucson, Arizona 85724*

Studies of the Treacher-Collins syndrome have emphasized hearing and surgical considerations. Although craniofacial anomalies have been associated with respiratory disorders in infancy, the presence of such problems in older children has not been emphasized. An eight-year-old with Treacher-Collins syndrome presented a history of recent behavioral problems at home, poor attention span and performance in school, daytime somnolence, and sleep apnea with relatively long periods of chest movement but no airflow. He also had abnormal sleep behavior consisting of rocking to and fro on his hands and knees, often to such an extent that his nose became abraded. ICU monitoring with observation and recording of sleep patterns and sounds, and fluoroscopy of his upper airway utilizing cineradiography while asleep confirmed the diagnosis of obstructive sleep apnea. The patient subsequently underwent an orthognathic surgery consisting of insertion of rib bone grafts after anterior advancement of his mandible. This procedure resulted in disappearance of the obstructive sleep apnea and associated symptoms. Because of micrognathia, patients with Treacher-Collins syndrome are at high risk for developing obstructive sleep apnea. Surgical correction of their deformities can result in improvement in cosmetic appearance as well as in resolution of the obstructive episodes with improvement in performance and behavior.

Mandibulo-facial dysostosis or Treacher-Collins syndrome was first described in 1888. Since then, there have been many case reports (Fazen et al., 1967; Fernandez and Ronis, 1964; O'Connor and Conway, 1950; Rogers, 1964; Rovin et al., 1964) which have emphasized the unusual facial features, hearing loss, and types of surgery for the facial problems, but none has discussed the possibility of obstructive sleep apnea in these individuals. For many years, children with this syndrome were considered to be retarded until it was discovered that they had marked hearing deficits which accounted for their delayed development. Obstructive sleep apnea has recently been described (Guilleminault et al., 1976) in children who apparently have learning difficulties, personality problems, daytime somnolence, and other manifestations which may be misinterpreted as learning disabilities or mental retardation secondary to other defects. Recent reports of sleep apnea (Phillipson, 1979; Cogswell and Easton, 1974; Lapidut and Ben-Hur, 1975) have described this entity in *infants* with certain craniofacial anomalies such as the Pierre-Robin syndrome, but no mention has been made of sleep apnea in Treacher-Collins syndrome. In addition, sleep apnea in older children with craniofacial abnormalities has not been described extensively. The case presented here highlights the fact that sleep apnea may occur with Treacher-Collins syndrome and emphasizes the importance of considering the possibility of sleep apnea in older children with craniofacial anomalies associated with personality and learning problems.

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The authors are affiliated with the Departments of Pediatrics, Family Medicine, Otolaryngology and Radiology, Arizona Health Sciences Center, Tucson, Arizona.

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## Case Report

An eight-year old white male (Figure 1) was first seen for evaluation of asthma. This patient had a diagnosis of Treacher-Collins syndrome since birth but had not undergone any reconstructive surgery. He wore bilateral hearing aids because of a severe conductive hearing loss, secondary to abnormalities of middle ear structures. Almost from birth, the child had been a noisy breather, especially at night, as evaluated by his mother. His snoring became so loud that, over the previous few months, his parents had to close their door at night. He had been observed to be extremely restless during sleep, and his mother complained that he fell asleep frequently during the day. His mother had noticed that the patient frequently slept on his hands and knees, often rocking to and fro and abrading his nose in the process. She had noticed several episodes where his chest appeared to be moving without any apparent exchange of air. Several months prior to the first clinic visit, reports of poor performance at school became frequent. He had also become a behavior problem at home. There was no enuresis.

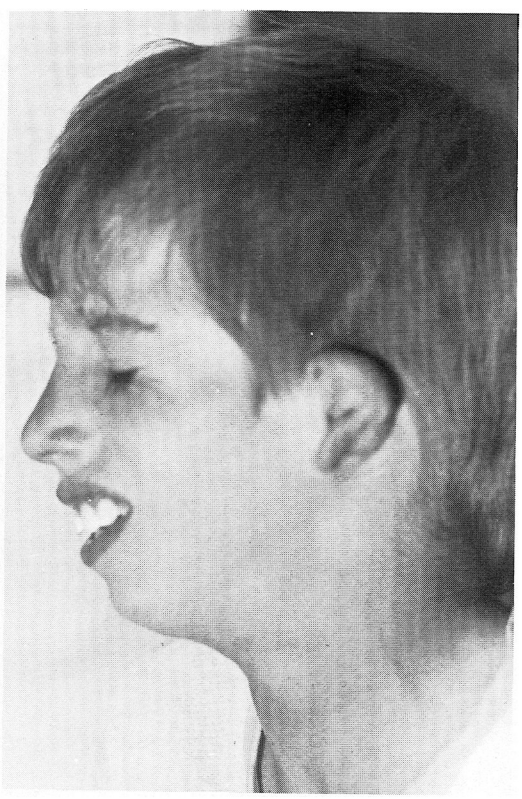

FIGURE 1. A pre-surgical side view of the patient, showing the small, receded mandible plus other features characteristic of the Treacher-Collins Syndrome. Nasal abrasion, acquired during sleep, is visible.

Because of this history, the patient was admitted to the Pediatric Intensive Care Unit at the Arizona Health Sciences Center for evaluation.

Initial physical examination revealed a cooperative eight-year-old boy, who was small for age. He had the classic findings of Treacher-Collins syndrome including antimongoloid shaped eyes, hypoplasia of the facial bones especially the maxilla and zygomatic bone with resultant micrognathia, extremely narrow nares, and malformed auricles. He wore a binaural hearing aid. Examination of the oropharynx showed tonsils of normal size with no evidence of obstruction. The adenoids were not easily visualized. The remainder of the physical examination, including that of the cardio-respiratory system, was normal. His blood pressure was 110/80 mmHg; respiratory rate was 24 breaths/min., and pulse was 120 beats/min. Initial laboratory results revealed a hemoglobin of 16.1 gms % and hematocrit of 45%. His chest radiograph and electrocardiogram were normal. Radiographs of the neck did not show any prominence of tonsillar or adenoidal tissue.

Sleep monitoring revealed obstructive apneic episodes often lasting from 30 to 35 seconds and occurring repeatedly throughout the night. During these apneic episodes, chest movement was noted in the absence of air flow. In addition, the patient slept in multiple positions ranging from sitting up to a knee-chest position. Tape recordings of his breathing revealed loud snoring with periods of complete silence lasting up to 30 or 35 seconds followed by a very loud snort and resumption of his previous breathing pattern. On a subsequent day, the patient was kept awake for a prolonged period of time and allowed to fall asleep (without sedation) on the cineradiography table in the Radiology Department. Cross-table cineradiographs were then obtained of the upper airway and pharyngeal structures during sleep. Within minutes of falling asleep, the posterior pharynx began to narrow with complete obstruction following (Figure 2). This occurred as the posterior

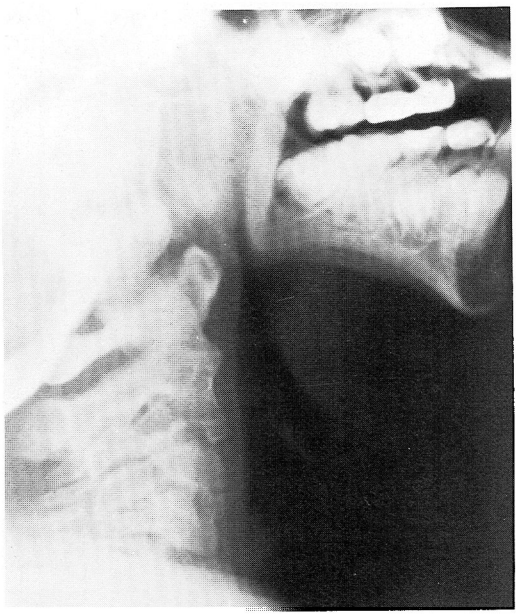

FIGURE 2. A preoperative lateral radiograph of the neck obtained with the patient sitting shows a patent airway.

aspect of the tongue relaxed and fell back occluding the airway. There was no evidence of collapse of the posterior pharyngeal wall. The obstruction would be complete for 20 to 25 seconds although respiratory efforts were observed. The obstruction was broken as the patient made a loud snorting noise, lifting the tongue forward and out of the posterior pharynx. This process was observed several times over a 30-minute period. The patient was then placed in the sitting position. The upper airway was again observed under cineography. Although the upper airway would narrow intermittently, complete obstruction was not observed in the sitting position (Figure 3). Lateral radiographs of the neck obtained with the patient awake in both the sitting and supine positions failed to demonstrate obstruction of the posterior pharynx. The findings on the cineradiographs indicated that the obstruction occurred because the tongue fell posteriorly into the oropharynx and obstructed the airway. For this reason, an orthognathic procedure for advancement of the mandible rather than a tonsillectomy and/or adenoidectomy was done. The surgical procedure of choice would have been a sagittal split of the mandible. However, because of the patient's religion (Jehovah's Witness), it was decided that better control of blood loss could be achieved by an extraoral procedure. The left anterior seventh and eighth ribs were removed. A Risdon approach was then done on the right and left sides of the mandible. The mandible was sectioned using a C-osteotomy with a vertical step at the inferior border of the mandible. A preformed acrylic splint was then fitted in the mouth. The patient was placed in intermaxillary fixation, and the rib grafts were then cut, fit to place, and wired. Total estimated blood loss was 358 cc, which was estimated to be about 1/4 of the patient's total blood volume. This was replaced with Ringer's Lactate, and no blood was given. The patient was kept in intermaxillary fixation for three months. He healed well without any post-operative problems.

Following surgery (Figure 4), there was almost total disappearance of the obstructive sleep apneic episodes; his breathing during sleep became relatively quiet; sleep occurred without the assumption of peculiar positions; daytime somnolence disappeared; and his behavior improved. Repeat sleep cineradiographs done six months after surgery failed

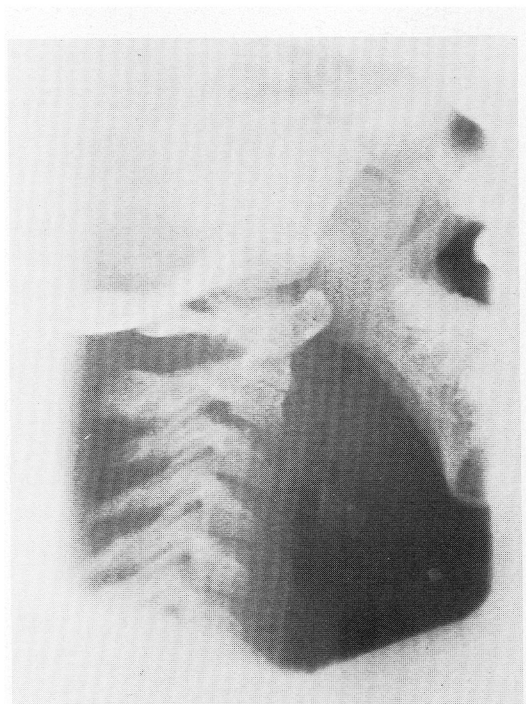

FIGURE 3. Sleeping supine lateral radiograph of the neck (taken from the cineradiographs) shows airway occlusion prior to surgery.

to reveal any episodes of upper airway obstruction and apnea. The findings noted on the initial study were now absent (Figure 5). His asthma was successfully treated with oral theophylline on a regular basis with intermittent need for inhaled beta sympathomimetic drugs.

## Discussion

Micrognathia associated with obstructive sleep episodes has been described in adult patients with acquired micrognathia (Conway, et al., 1977). Upper airway obstruction secondary to craniofacial anomalies in the pediatric age range has been described almost exclusively in neonates with the Pierre-Robin syndrome. Chronic upper airway obstruction with development of cor pulmonale and/or obstructive sleep apnea in older children has been due, in nearly all reported cases, to enlarged tonsils and adenoids or to relaxation of the tongue and pharyngeal walls (Ainger, 1968; Djalilion et al., 1975; Goodman et al., 1976; Levin et al., 1975; Luke et al., 1966; Menaske et al., 1965). The case reported here highlights the importance of investigating for chronic upper airway obstruction and sleep apnea in older children with craniofacial abnormalities. Because children with Treacher-Collins syndrome have severe hearing deficits, one may attribute learning problems as well as personality disorders to the hearing deficit and to other associated problems. Therefore, any child with craniofacial anomalies such as those associated with the Treacher-Collins syndrome in combination with poor school performance or behavioral disturbances or both should be studied for obstructive sleep apnea.

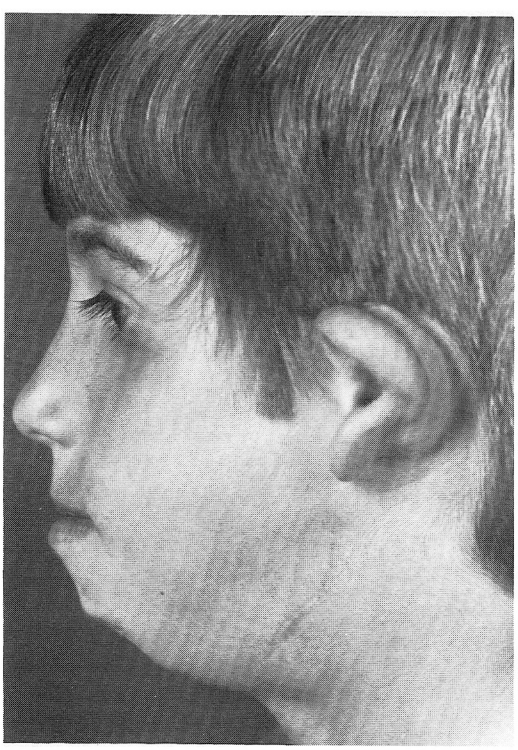

FIGURE 4. Postsurgical side view of the patient. Compare to Figure 1.

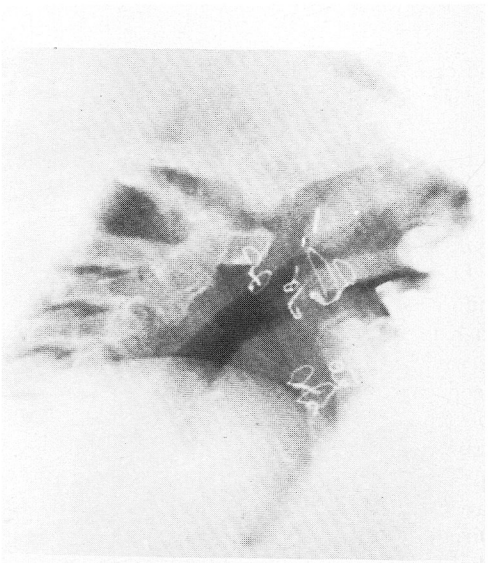

FIGURE 5. Post-surgery sleeping supine lateral radiograph of the neck (taken from the cineradiographs) shows a widely patent airway. No episodes of obstructions, as shown in Figure 3, were observed.

The history is very important in attempting to identify individuals who may suffer from sleep apnea. The patient, or more frequently the family, might complain of any of the following: excessive daytime sleepiness, nocturnal insomnia, noisy snoring, abnormal motor activity during sleep, intellectual and personality changes, morning headache, and enuresis. Clinical findings in the more severely affected individuals may include systemic hypertension, cor pulmonale, congestive heart failure, polycythemia, and hypoxia (especially while asleep), a very sleepy child, elevated blood pressure, a loud pulmonic component of the second heart sound, and an active right precordium with a heave, indicative of increased pulmonary artery pressure. However,

the episodes of obstructive sleep apnea may be of such short duration that the cardiopulmonary exam may be normal as was the case in our patient. Therefore, sleep apnea should be suspected even if the older child does not show signs of cor pulmonale or pulmonary hypertension.

Documentation of obstructive sleep apnea requires hospitalization and close monitoring with recording of breathing patterns as well as pulse. Examination must be done during sleep, and a variety of modalities can be used to evaluate sleep state, degree of airway obstruction, impairment of ventilation, development of hypoxemia and hypercapnia, and cardiac function during the apneic episodes. Direct visualization of the upper airway and pharyngeal structures during sleep by cineradiography provides conclusive evidence of obstruction. Since certain drugs may have a direct effect on the muscular tone of the structures of the pharynx, it is best to try to obtain the radiographs and cines while the patient is asleep without sedation.

Episodes of sleep apnea (defined here as a cessation of air flow across the nose or mouth for more than 15 seconds) may occur as frequently as 500 times a night in severely affected individuals. There are two major forms of sleep apnea that can exist alone or in combination. The first form, obstructive sleep apnea, is apnea that occurs despite continued or even enhanced respiratory efforts. This usually occurs secondary to pharyngeal or upper airway obstruction. The second form is central sleep apnea in which there is total lack of respiratory effort. The two forms can occur together. When they do, the initial apneic episode usually starts as central apnea, which is then followed by an obstructive component. The mechanism(s) by which the obstructive process may develop in patients such as the one described here are still unclear, but several theories have been advanced. General hypotonia of the tongue may allow it to fall backward, occluding the airway. If the pharyngeal space is small (as in patients with craniofacial anomalies), only a small degree of backward movement of the tongue may lead to airway obstruction. Severe nasal obstruction may also be present. Each or both of these mechanisms may cause the patient to produce a marked negative pressure within the pharynx during inspiration. This negative pressure would then enhance posterior movement of the tongue, collapse of the pharyngeal soft tissues, or both thereby contributing to the obstruction (Mallory and Paradise, 1979). In addition, some patients may not have a normal level of genioglossus activity during sleep. This allows the tongue to fall backward and obstruct the airway. The acute upper airway obstruction is overcome when hypoxia and hypercapnia develop and thereby stimulate the respiratory centers. Respiration then occurs with the voluntary lifting of the tongue. This repetitive arousal pattern is thought to be one possible mechanism for the behavior changes as well as for the daytime somnolence. Systemic abnormalities such as hypertension, heart failure, and pulmonary hypertension are thought to be secondary to the chronic hypoxemia and hypercapnia which occur predominantly at night.

Treatment of the patient with sleep apnea will vary depending on the type of apnea present. Central sleep apnea has been successfully treated with a number of drugs, most commonly theophylline. The treatment of obstructive apnea is directed at the correction of the physical problem. Initially, establishment of a nasopharyngeal airway can often provide dramatic temporary relief. Movement of the mandible anteriorly, as in the patient described, may relieve the symptoms. Occasionally a tracheostomy may be required. The dramatic improvement in symptoms and the disappearance of airway obstruction following surgery in this patient is further support that mandibular advancement was the treatment of choice.

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