

SLEEP APNEA AND STICKLER SYNDROME

Use of oral appliance therapy for sleep apnea benefited a male patient with respiratory distress syndrome and obstructive sleep apnea.

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A male patient born in 1979 with respiratory distress syndrome had surgery performed in the first month of life to repair a patent ductus arteriosus. From 1980 to 1983, five or six palate repairs were performed. In 1984, a pharyngeal flap removal was done, followed by an emergency reversal. In 1984, a Le Fort I surgery was performed to correct a retrognathic jaw, and a tonsillectomy and adenoidectomy were performed. Between 1986 and 1993, tubes were placed in the patient's ears six times for chronic otitis media. In 1996, another Le Fort I procedure was performed to correct sleep apnea. This surgery was a failure that made the condition worse.

Severe myopia and astigmatism were present, and the patient wore corrective lenses beginning at 18 months of age. The patient had significant speech problems, requiring therapy from ages 3 to 15 years. He also had corrective orthodontic work performed.

He was diagnosed as having Pierre Robin syndrome, which is named for the French physician who identified the main features of the condition. The syndrome is rare, with estimates of its incidence ranging from one in 8,000 to one in 30,000 people. The features of Pierre Robin syndrome include micrognathia; glossoptosis (a tendency for the base of the tongue to ball up and fall backward into the throat, causing obstruction and, therefore, breathing difficulties); and a cleft palate, or a high-arched palate that is not cleft. The jaw bone continues to grow during childhood and will usually reach full size by adulthood.

Later, the patient was diagnosed with Stickler syndrome,¹⁻¹¹ an autosomal-dominant condition with genetic heterogeneity in which some of the procollagen genes are implicated (frequently on chromosomes 6 and 12). It affects one in 10,000 people and is associated with connective-tissue problems, visual problems such as myopia, hearing loss, early-onset arthritis, cleft palate, and heart murmur. The patient had a severe speech defect and some craniofacial deformity.¹⁻¹¹

To help him cope with his many physical and mental concerns, the patient saw a psychiatrist at Sick Children's Hospital, Toronto, Ontario, until 1988 and then started seeing another psychiatrist weekly. Major issues included coping with his physical and medical problems, the separation of his parents when he was 8 years old, and the death of his stepfather in 1997; however, the patient was not depressed. He tended to have a positive outlook and a good sense of humor, and he socialized with friends.

CASE HISTORY

The patient had a history of sleep apnea beginning in early childhood, resulting in fatigue and continuing sleepiness. He was diagnosed with obstructive sleep apnea (OSA) in 1984, and it had probably been present since he was 2 years old. The patient had undergone more than 20 overnight sleep studies.

After the patient's second (and unsuccessful) Le Fort I

procedure in 1996, subsequent strategies tried included continuous positive airway pressure (CPAP) therapy, which he was unable to tolerate due to jaw deformities. Various antidepressants were prescribed, including imipramine (which was of some benefit for 10 months), desipramine (which was of no benefit), and nefazodone at 200 mg twice daily, which he continues to take. In 1997, asthma was diagnosed by a respirologist at Toronto Western Hospital. A diagnostic sleep study showed moderately severe apnea, with an apnea-hypopnea index (AHI) of 40.

In 1998, the patient discovered that sleeping in a reclining chair stopped his apnea. This was effective for 10 months, as confirmed by polysomnography, but was discontinued because severe joint pain that had developed in the patient's back and neck completely debilitated him.

In March 1999, a respirologist at Toronto Western Hospital referred the patient to a dentist for oral appliance therapy for sleep apnea. On April 8, 1999, a mandibular repositioning appliance was inserted.^{12,13} There were certain difficulties in appliance therapy; the patient's small teeth caused problems in retention of the oral appliance, for example. A specific appliance was chosen to handle this problem. The small range of motion of the lower jaw could have posed a problem, but the dentist expected range of motion to improve significantly with use of the oral appliance. The dentist wanted to extend the mandible as far forward as possible, as quickly as possible, to improve the patient's OSA condition.

RESULTS

The problem of a small mandibular range of motion did not appear to diminish, but it did not prevent a positive result in this case. A sleep study with the dental device was done on April 9, 1999 (the day after insertion); there was, however, some confusion between the technicians and the patient as to its correct adjustment, so the study's results were inconclusive. The dentist examined the patient on May 31, 1999. The patient was used to the appliance, and it had beneficial results.

A new sleep study was performed with the appliance in place in January 2000. It showed an AHI of less than 5, with no fragmentation or oxygen desaturations. This indicated that the oral appliance stopped the apnea problem as long as it was used. The patient used it nightly.

A multiple sleep latency test was performed the following day. The patient fell asleep during three of the four naps within about 5 minutes. There were no rapid-eye-movement intrusions. This showed that a problem in addition to apnea was still causing the patient to be sleepy and fatigued.

DIAGNOSIS

A new consulting sleep psychiatrist was seen for help with

fatigue. He noted that the dental appliance made it unnecessary for the patient to sleep in a reclining chair and that he fell asleep in 15 minutes and slept deeply through the night. When he awoke, he did not generally feel particularly bad and did not describe being excessively fatigued. It was believed that the patient was coping much better and was reasonably happy with his social life.

This sleep consultant was very impressed by the beneficial effects of the dental appliance, which had substantially improved his sleep-wake condition. The patient continued to require close monitoring by the dentist for adjustments of the appliance. He continued to require supportive psychotherapy with his regular psychiatrist to help consolidate the gains that had been made over the previous year. The question was raised whether he needed to continue to rely on nefazodone, and whether the medication might contribute to his lethargy. This specialist did not believe that further intervention was necessary from the Center for Sleep and Chronobiology (CSC), Toronto, and did not suggest a follow-up appointment. The patient was deferred to his dentist and regular psychiatrist, who had been providing exemplary care; if they believed the CSC services were required, they would be pleased to see the patient.

As of February 4, 2000, the patient still had a significant problem with fatigue and daytime sleepiness. He could not work or attend school, and his mental stamina was impaired. He slept 9 to 13 hours per day and would wake between 10 AM and 7 PM. The patient had shortness of breath at times. Sometimes he would be able to carry out a daily routine.

The patient used the oral device nightly for 11 months. He was able to think in the morning, with his best times being between 10 AM and 3 PM. He was disappointed that he could not hold a job or go to school, but he kept himself motivated and liked to use a computer.

Sleep times varied over a 24-hour period and were very erratic. As a result, the patient would be very tired during the day. He found some relief through the use of various dietary supplements. He frequently experienced pain in his hips, knees, right shoulder, and neck. He was a beneficiary of the Ontario Disability Service Plan, and caregiver-burden issues were experienced by his mother and sister.

The patient was seen by a dentist in the autumn of 2000 and informed him that the respirologist believed he needed different medication for asthma. This medication included taking 125 mg of fluticasone twice daily, 2 puffs of mometasone furoate monohydrate nasal spray in each nostril once daily, and albuterol as required, which improved the patient's alertness significantly, and allowed him to feel well enough to consider a job search. The sleep consultant prescribed 50 mg of sertraline hydrochloride at bedtime to help him get to sleep, 100 mg of modafinil twice daily to help him arouse, and he continued to use the oral appliance to correct his apnea. As of May 15, 2001, the patient had much less fatigue and sleepiness and was holding a part-time job and taking computer courses.

On March 20, 2002, the patient received a new mandibular repositioning device, as the old one was breaking. The patient was still doing very well with his oral appliance and asthma medication. It was decided that apnea was no longer a problem, as long as the appliance was effective.¹⁴ Using the oral appliance, the patient was able to function much better during the day and was able to sleep in a normal bed at night, avoiding the neck and back pain he had experienced with postural sleep therapy. His oral appliance therapy will be continuously monitored.

CONCLUSION

The importance of an oral appliance for moderately-severe apnea in a patient for whom CPAP would not work was demonstrated.^{15,16} Another surprising result was that two Le Fort I surgical mandibular advancement surgeries were performed¹⁷ to open the airway, but did not improve the apnea, whereas the oral appliance (which is presumed to open the airway in a similar manner by advancing the jaw nonsurgically) was effective.

The multiple problems experienced by this patient show the extreme importance of the input of various specialties. The sleep rounds, at which this patient was present as the subject, was presented at the Center for Sleep and Chronobiology on February 4, 2002. This was the third time this patient was presented as the subject of the rounds, which enabled wide-ranging and fast access to information from psychiatrists, respirologists, otorhinolaryngologists, rheumatologists, orthopedists, and dentists. **SR**

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