Down Syndrome and Sleep-Disordered Breathing: The Dentist’s Role
H. Barry Waldman, Faysal M. Hasan and Steven Perlman
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Down syndrome and sleep-disordered breathing
The dentist’s role

H. Barry Waldman, DDS, MPH, PhD; Faysal M. Hasan, MD; Steven Perlman, DDS, MScD

ABSTRACT

Background. Sleep apnea is a common sleep disorder characterized by sleep fragmentation, oxygen desaturation and daytime somnolence. It is common in people with Down syndrome, occurring in at least one-half of this population.

Methods. The authors review the clinical presentation of obstructive sleep apnea and its association with Down syndrome.

Results. The diagnosis and treatment of sleep-disordered breathing in the Down syndrome population reinforce the important role that dentists play in recognizing its association with comorbidities and daily cognitive and behavioral function.

Conclusions. Sleep abnormalities in children with Down syndrome often are not cured by surgical procedures. These patients may not tolerate continuous positive airway pressure treatment, but they should benefit from the use of oral appliances.

Clinical Implications. The dentist has an important role in enhancing the quality of life of patients with Down syndrome, especially in the recognition and treatment of sleep-disordered breathing.

Key Words. Down syndrome; obstructive sleep apnea; dental care for disabled; dental devices.

Association between DS and sleep-disordered breathing has gone unrecognized by parents and health care practitioners, which contributes to learning and behavioral problems.

OSA is characterized by episodic obstruction of the upper airway during sleep, resulting in hypoxemia and frequent arousals; it is associated with symptoms such as snoring and daytime hypersomnolence. OSA has a high prevalence in the U.S. adult population, affecting at least 4 percent of men and 2 percent of women in the middle-aged workforce. The treatment approach for children with sleep-disordered breathing has focused on surgical options such as tonsillectomy and adenoidectomy, but OSA in patients with DS often persists even after surgery.

The “gold standard” for OSA treatment is the use of a tight face mask and continuous positive airway pressure (CPAP) breathing. However, patient compliance is a problem, and some clinicians recommend the use of oral appliances and other surgical approaches as an alternative therapy for less severe cases. These treatment approaches require the collaboration of health care providers in several disciplines, including pulmonology, neurology, psychiatry, otolaryngology and dentistry.

The practicing dentist and staff members are in a unique position to recognize sleep-disordered breathing and screen for it in this population with DS. They must have a high index of suspicion, recognize the maxillofacial and pharyngeal features of the disease, and be aware of the patient’s history of snoring and daytime somnolence. The dentist may help patients who have this condition by developing expertise in the use of dental appliances to treat OSA.

CLINICAL FEATURES OF DOWN SYNDROME AND ASSOCIATION WITH OBSTRUCTIVE SLEEP APNEA

More than 350,000 people in the United States have DS. Approximately 5,000 children are born with DS annually. The condition is not related to race, nationality, religion or socioeconomic status, but its risk increases with maternal age. It occurs in one of 100 births in women older than 40 years. Advances in medical treatments have greatly improved the quality of life and life expectancy of children with DS. For example, repair of congenital cardiac defects now is routine, and advocates for patients with OSA have suggested that children with DS should not be deprived of advanced medical procedures.

Only a few decades ago, the mean life expectancy for people with DS was less than 20 years; today, the majority of people with DS live past the age of 55 years. The median age of survival for people with DS in the United States has increased from 2 years in 1968 to 50 years for whites and 25 years for African-Americans. In general, people with DS now are deinstitutionalized and have become more functional in the community and dependent on health care practitioners in their neighborhoods. Given this population’s improved longevity and quality of life, the recognition and treatment of OSA become vital as they may have a significant impact on morbidity, brain function and daytime performance.

Children with DS have multiple predisposing factors for the development of OSA, primarily because of their unique facial and upper-airway features. These factors include midfacial and mandibular hypoplasia, an abnormally small hypopharynx, encroaching tonsils and adenoids, and generalized pharyngeal muscle hypotonia with collapse of the upper airway during sleep. The tongue may protrude and appear too large, but true macroglossia is rare; rather, the oral cavity is smaller because of a low and narrow palate and underdevelopment of the midface.

Children with DS are recognized clinically by the finding of a “flat” face. They also may have an upward slant to their eyes, a short neck, abnormally shaped ears and white spots on the irises of their eyes. Children with DS typically have small hands and feet, with a deep crease in the palm of the hand, and poor muscle tone. They also have a small nasal airway, and, thus, chronic mouth breathing is common. This, in turn, leads to dry mouth and tongue and lip fissures. In addition, a compromised immune response in patients with DS can lead to severe periodontal disease. However, the prevalence of caries has been low. This may be due to a supervised or institutionalized diet or the delayed eruption of, and increased spacing between, teeth.

Patients with DS are predisposed to serious comorbidities, including congenital heart disease,
auditory and visual abnormalities, congenital gastrointestinal defects resulting in bowel obstruction and skeletal malformations. They also are prone to celiac disease, leukemia, thyroid dysfunction and premature dementia, appearing as early as 40 years of age. Moreover, these patients often are obese and have an abnormal immune system that results in an increased incidence of lower-respiratory-tract infections and pulmonary secretions.¹

OSA in children with DS often remains unsuspected and thus underdiagnosed. Many of the sequelaes of OSA—such as pulmonary hypertension, delayed development and behavioral abnormalities—also are commonly associated with DS. In general, parents’ descriptions of their children’s medical histories alone cannot be a reliable basis for screening children for OSA, because parents often are unaware of the symptoms. Parents invariably underestimate the severity of their children’s sleep disturbance. In one study, the parents of only 32 percent of children with documented OSA reported having suspicions of a sleep disturbance.³ In another study, sleep study findings were abnormal in 100 percent of children with DS.¹⁴ Inaccurate reporting may be more common to parents of children with DS, because questionnaires from parents of children without DS showed better correlation with polysomnographic findings.¹⁴ Given the high prevalence of OSA in children with DS, clinicians should have a high clinical suspicion of this diagnosis in these children.²⁰

OSA also is associated with neurocognitive abnormalities. Nocturnal oxygen desaturation and excessive arousals associated with sleep-disordered breathing are correlated with lower intelligence quotient performance testing and increased behavioral abnormalities.¹³ Chervin and colleagues²⁹ linked excessive sleep arousals and sleep deprivation with daytime hypersomnia, lack of energy and lack of motivation. Sleep fragmentation and deprivation may affect daytime performance and could exacerbate learning disorders. Clinicians and parents may overlook the resultant behavior and learning abnormalities, assuming that they are caused by limited abilities commonly assigned to children with DS.

RECOGNITION OF OBSTRUCTIVE SLEEP APNEA

Although traditional risk factors for patients with sleep apnea include obesity, male sex and middle age, OSA affects both sexes of all ages and people of ideal weight. Sleep apnea is two to three times more prevalent in men than in women, and this may be a reflection of sex differences in body fat distribution in the upper airway, as well as hormonal influences, or underdiagnosis, in women. A higher prevalence of sleep apnea has been described in patients with hypothyroidism and acromegaly, as well as in patients with neuromuscular disease that affects upper-airway muscle function.¹¹,¹²,³⁰ Risk factors include menopausal status and African, Chinese, Indian, Mexican and Polynesian ancestry. OSA, if untreated, is associated with major consequences that include systemic and pulmonary hypertension, myocardial infarction, left ventricular dysfunction, congestive heart failure, cardiac dysrhythmias, insulin resistance and stroke.¹¹,¹²,³⁰

OSA has been described in patients with facial and maxillary abnormalities such as retrognaethia that lead to posterior displacement of the genioglossus muscle, resulting in a narrowed hypopharynx. Airflow obstruction, despite the patient’s continued effort to breathe, results in apneas or hypopneas and cyclical oxygen desaturation (Table 1).³¹-³³ This results in stimulated breathing against a compromised airway, and the patient is awakened by repeated arousals.

Screening patients for OSA by the dentist and staff members is not cumbersome. It is important for them to ask patients and their parents about symptoms of snoring during sleep, any witnessed cessation of breathing, feeling tired or unrefreshed on awakening, and daytime hypersomnia or fatigue (Box). Clinicians can quantify patients’ daytime hypersomnia by administering the Epworth Sleepiness Scale³⁴ to their parents or guardians (Table 2, page 311). (A total score greater than 10 suggests excessive daytime sleepiness.) Physical examination findings should raise concerns about OSA if the oropharynx is compromised or crowded by a low soft palate, a large tongue or enlarged tonsils, and an increased neck circumference.

Clinicians usually confirm the diagnosis of OSA via all-night polysomnography, a continuous recording of electrophysiological variables during sleep. The test monitors brain electrical activity, eye and chin muscle tone, nasal and oral airflow, respiratory effort, heart electrical activity (via one-lead electrocardiography) and oxygen saturation to assess their physiological impact. Calf muscle tone also is recorded via electromyography
to monitor for periodic limb movements, which can be another cause of sleep fragmentation.14 The clinician usually determines the severity of OSA on the basis of the apnea-hypopnea index (Table 3).

**ROLE FOR ORAL DEVICES**

Weight loss, strict avoidance of alcohol and good sleep habits are the first steps in the treatment of OSA. Good sleep habits include maintaining regular sleep hours, reserving a bedroom for sleeping only, going to bed when drowsy, leaving the bed-room if one cannot fall asleep or stay asleep, and waking at the same time regardless of the duration of sleep.35 Patients may need to lose 10 to 15 percent of their weight before they experience a significant decrease in OSA severity; however, few patients are able to lose that much weight and keep it off.36

Effective treatment of OSA in children usually consists of tonsillectomy and adenoidectomy.27 However, this surgery may not be curative in children with DS because of the facial and muscular abnormalities that are peculiar to this population. More than 50 percent of children with DS continue to have evidence of upper-airway obstruction during sleep after a tonsillectomy and an adenoidectomy.38 Thus, a postoperative sleep study is recommended to determine if the treatment has been effective.38

**CPAP.** If a tonsillectomy does not resolve the patient’s OSA, Dyken and colleagues14 recommend CPAP as the preferred mode of therapy. The CPAP appliance works by splinting the upper airway and preventing the collapse of the soft tissue behind the palate and tongue. CPAP has been shown to eliminate apneas and hypopneas, decrease the number of nocturnal arousals and normalize oxygen saturation.39

Unfortunately, noncompliance is a major problem with CPAP. Shott40 reported that only one-half of patients use the appliance on a regular basis, and then for only four to five hours per night. The use of heated humidity and an improved interface between the mask and the face (such as the use of nasal pillows and gel masks), as well as clinicians’ providing increased education about the need for sleep and making weekly telephone calls to parents or guardians, appear to improve compliance. Unfortunately, CPAP is not well-tolerated by children or by those with developmental disorders such as DS.41

**Oral appliances.** Oral appliances are used increasingly in the treatment of mild-to-moderate OSA or in patients who cannot tolerate CPAP.41,42 They generally fall into two broad categories: tongue-retaining devices and mandibular advancement devices. By repositioning the lower jaw and thus the tongue, the oral appliance keeps the pharyngeal airway open. Tongue retainers

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**TABLE 1**

<table>
<thead>
<tr>
<th>TERM</th>
<th>DEFINITION</th>
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<tbody>
<tr>
<td>Apnea</td>
<td>Absence of breath for 10 seconds or longer</td>
</tr>
<tr>
<td>Apnea-Hypopnea Index</td>
<td>Number of apneas, hypopneas or both during one hour of sleep</td>
</tr>
<tr>
<td>Central Apnea</td>
<td>Observed cessation of breathing without chest wall motion</td>
</tr>
<tr>
<td>Hypopnea</td>
<td>Decreased amplitude of tidal breath of more than 40%, accompanied by a 4% decline in oxygen saturation</td>
</tr>
<tr>
<td>Nasal Continuous Positive Airway Pressure (CPAP)</td>
<td>CPAP breathing by using a tight, sealed facial mask</td>
</tr>
<tr>
<td>Obstructive Apnea</td>
<td>Observed cessation of breathing despite chest wall effort</td>
</tr>
<tr>
<td>Obstructive Sleep Apnea</td>
<td>Five or more apneas or hypopneas observed per hour during sleep (see Table 3 for determination of severity)</td>
</tr>
<tr>
<td>Oxygen Desaturation</td>
<td>Drop in saturation below 90% or more than 4% from baseline, according to pulse oximetry</td>
</tr>
<tr>
<td>Respiratory Disturbance Index</td>
<td>Number of apneas and hypopneas resulting in arousals, irrespective of degree of desaturation, during one hour of sleep</td>
</tr>
</tbody>
</table>

* Sources: Stedman’s Medical Dictionary,31 Taber’s Cyclopedic Medical Dictionary,32 Medical Dictionary Online.33
are used less commonly and are more cumbersome, but they tend to be an effective appliance. The tongue retainer has a suction cup that attaches to the front of the tongue, pulling it forward to prevent the tongue from collapsing. However, people with DS may find this type of appliance particularly difficult to tolerate.\(^4\)

The most effective oral appliances for patients with DS are mandibular advancement devices that fit both the maxillary and mandibular teeth, similar to an orthodontic retainer or an athletic mouthguard. The dentist can set them arbitrarily to protrude the mandible, or they can be adjusted in increments to achieve the optimal balance in reducing the severity of OSA without causing temporomandibular discomfort.

Mandibular advancement devices have been shown to reduce the number of apneas and hypopneas, improve oxygen desaturation and improve overnight sleep quality.\(^4\) They work best for patients with primary snoring and in mild-to-moderate OSA cases.\(^4\) Numerous studies\(^4\) support oral appliance therapy for people with sleep apnea. Overall data indicate a 50 to 60 percent reduction in the respiratory disturbance index of patients with OSA, with a patient acceptance rate of 75 percent.\(^4\)

Considerable data confirm the efficacy of dental appliances for OSAs, but they are successful in only one-half of patients.\(^4\) In general, oral appliances are less effective than CPAP therapy, but they have fewer side effects, and patients prefer them to CPAP according to comparisons in some studies.\(^4\)

More than 50 dental devices are on the market for sleep apnea.\(^4\) Oral appliances are portable and require no cumbersome headgear, nasal mask or electric power. However, the patient must have both upper and lower teeth. The literature contains reports of soft-tissue irritation and excessive salivation, as well as discomfort of the masseter muscles, temporomandibular joints or both, associated with these appliances.\(^4\)

Patients’ responses to oral appliances may be affected by their pharyngeal anatomy. A comparison of the risks and benefits of oral appliance therapy with those of other treatment modalities suggests that oral appliances are a viable alternative to CPAP.\(^4\)

### TABLE 2

<table>
<thead>
<tr>
<th>Epworth Sleepiness Scale.*</th>
</tr>
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<tbody>
<tr>
<td>• How likely are you to doze off or fall asleep in the following situations, in contrast to feeling just tired?</td>
</tr>
<tr>
<td>• This refers to your usual way of life in recent times.</td>
</tr>
<tr>
<td>• Even if you have not done some of these things recently, try to work out how they would have affected you.</td>
</tr>
<tr>
<td>Use the following scale to choose the most appropriate number for each situation:</td>
</tr>
<tr>
<td>0 = would never doze</td>
</tr>
<tr>
<td>1 = slight chance of dozing</td>
</tr>
<tr>
<td>2 = moderate chance of dozing</td>
</tr>
<tr>
<td>3 = high chance of dozing</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>SITUATION</th>
<th>CHANCE OF DOZING (0-3)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sitting and reading</td>
<td>—</td>
</tr>
<tr>
<td>Watching TV</td>
<td>—</td>
</tr>
<tr>
<td>Sitting inactive in a public place (such as a theater)</td>
<td>—</td>
</tr>
<tr>
<td>Passenger in a car for one hour without a break</td>
<td>—</td>
</tr>
<tr>
<td>Lying down to rest in the afternoon</td>
<td>—</td>
</tr>
<tr>
<td>Sitting and talking to someone</td>
<td>—</td>
</tr>
<tr>
<td>Sitting quietly after a lunch without alcohol</td>
<td>—</td>
</tr>
<tr>
<td>In a car, stopped for a few minutes in traffic</td>
<td>—</td>
</tr>
<tr>
<td>TOTAL SCORE</td>
<td>—</td>
</tr>
</tbody>
</table>

* Adapted with permission of Dr. Murray Johns from Epworth Sleepiness Scale.\(^4\)

### TABLE 3

<table>
<thead>
<tr>
<th>Severity</th>
<th>Apnea-Hypopnea Index (AHI)</th>
<th>Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal</td>
<td>&lt; 1 for children, &lt; 5 for adults</td>
<td>Conservative measures, sleep hygiene(^4)</td>
</tr>
<tr>
<td>Mild</td>
<td>5-14</td>
<td>Oral appliances, surgery, CPAP</td>
</tr>
<tr>
<td>Moderate</td>
<td>15-30</td>
<td>CPAP, oral appliances</td>
</tr>
<tr>
<td>Severe</td>
<td>&gt; 30</td>
<td>CPAP, tracheostomy</td>
</tr>
</tbody>
</table>

* Some patients with a normal AHI have increased arousals and daytime sleepiness as a result of increased respiratory event–related arousals. These patients have the upper-airway resistant syndrome and may require treatment with continuous positive airway pressure (CPAP).
† Maintenance of weight hygiene, avoidance of supine position, abstinence from alcohol within four hours of sleep and maintenance of nasal patency.

### CONCLUSION

For people with DS, the combined reality of deinstitutionalization, increased life spans and a
strong association with OSA creates opportunities for community dental practitioners to play an important role in their treatment. The dentist should recognize the impact of sleep impairment and the need for diagnostic testing to confirm suspicions when evaluating a patient’s breathing management. He or she should collaborate with a physician who is a sleep specialist and the patient’s primary physician when evaluating and treating patients with DS and OSA.

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