

Case Vignette: OSA and Autism Spectrum Disorder

Presenting Complaints

A 20-year-old African American male presented to his family physician along with his mother for some guidance on sleep hygiene. According to his mother, he had poor sleep habits, frequently went to bed after midnight and got up late in the morning, but she could not give specific times. She also noticed that he snored loudly and often. She added that while he did not take daytime naps, he did appear lethargic and sleepy many times during the late morning and early afternoons. The patient was not presently employed or in school.

Past History

The family physician was familiar with his past medical history, which was significant for severe ASD and seizure disorder, and he was mostly non-verbal. His mother reported that he took the following medications regularly: Quetiapinel, Carbamazepine extended release, Clonidine, and Fluoxetine. He had expressed a high food selectivity from a young age, favored chicken nuggets and 2 slices of bread with butter as the main meal and consumed less fruit and vegetables. In the past one year or so, he had doubled his intake, and this resulted in a 15-20 lb. weight gain. His mother denied the patient's use of caffeine and alcohol.

Physical Exam

During the clinical assessment, the patient appeared disinterested and was easily distracted. He made little eye contact either when listening and his verbal interaction was very limited. His answers to questions were minimal and his speech lacked normal prosody.

- Blood pressure and pulse: Within normal range
- BMI: 31 kg/m²
- Chest and abdomen clear

Testing

His BMI of 31 kg/m² and snoring at night raised concerns about potential sleep apnea and warranted further investigation. His family physician then asked the mom to complete the Epworth Sleepiness Scale (ESS) questionnaire to the best of her ability to assess her son's sleepiness.



The ESS subjectively assesses excessive daytime sleepiness by asking patients to rate their chance of dozing off from 0 (would never doze) to 3 (high chance of dozing) for 8 commonly encountered scenarios, with a total maximal score of 24.

Would never doze	Slight chance of dozing	Moderate chance of dozing	High chance of dozing	
0	•	0	0	Sitting and reading
0	0	•	0	Watching TV
0	•	0	0	Sitting inactive in a public place (e.g. cinema or in a meeting)
0	0	•	0	Being in a car for an hour as a passenger (without a break)
0	0	•	0	Lying down to rest in the afternoon (when possible)
0	•	0	0	Sitting and chatting to someone
0	•	0	0	Sitting quietly after lunch (not having had alcohol)
•	0	0	0	In a car when you stop in traffic for a few minutes
Calculate Score				
Your Score				
10				

Figure 1. Results of ESS questionnaire

The ESS score generated was 10/24 (Figure 1). ESS scores of 6-10 depicts a "higher normal daytime sleepiness" [About the ESS]

His family physician put in a referral for a sleep medicine specialist and ordered a sleep study.

An overnight sleep study identified 64 obstructive, one mixed, and 57 hypopneas. The Apnea-Hypopnea Index (AHI) was 12.4 for sleep in the supine position and 12.7 for sleep in the side position. The oxygen saturation averaged 95.9 percent.



Throughout much of the study, the patient's end tidal CO₂ count was above 55 torr, which is higher than the normal range of 35 to 45 torr. Ninety-eight spontaneous arousals (10.1 per hour) were noted. A diagnosis of mild OSA and hypercapnia was given.

Discussion of Treatment Plan

Based on these findings, the sleep medicine specialist recommended treatment for sleep apnea and presented the following options: dental appliance; oral surgery; and weight loss. Although CPAP therapy is the most common treatment for OSA, it was felt that the patient's limited understanding would prevent him from being able to wear a mask. His mother expressed interest in the dental appliance option and was referred to the nearby university's Dentistry Special Needs Clinic.

At the appointment, his mother provided interpretation of his symptoms, sleepiness, and fatigue level, given that his ASD made identification and interpretation of symptoms impossible in the clinical setting. A dental and occlusal exam was not possible. Class II molar and canine relationships and normal jaw mobility were inferred. His mother reported that he had no caries history.

During the next several weeks, the patient was gradually acclimatized to tolerate an intraoral object starting with a one piece 'boil and bite' mouthguard, followed by a two-piece 'boil and bite' mouthguards. Subsequently and with careful coaching, the patient was then asked to use a temporary but customized mandibular advancement splint to wear nightly. This also served as a trial device to evaluate the patient's tolerance of jaw advancement.

Four months after the initial appointment, the patient returned to the clinic for delivery of the temporary device. He adapted very well to it and wore it for five to six hours every night, during which time his mother reported much less intense snoring. The jaw advancement was increased from the initial 60% of maximum jaw protrusion (MJP) to 75% MJP to further reduce snoring. Eight months after the delivery of the temporary device, the patient was able to insert and remove it on his own. The appliance was then adjusted to protrude the mandible at 80% MJP. 2 months later, the final appliance was made to match the final advancement of the temporary appliance, which the patient tolerated. He was encouraged to chew sugar-free gum in the mornings to regain his natural bite after removing the appliance. His mother was instructed on care of the appliance. (Figure 2)



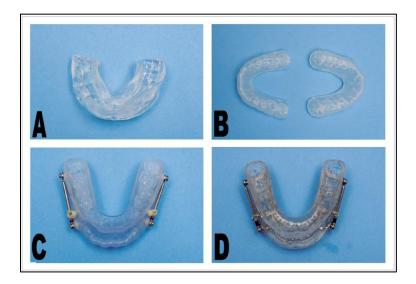


Figure 2: (A) One piece 'boil and bite' mouthguard. (B) Two-piece 'boil and bite' mouthguards. (C) Temporary mandibular advancement device. (D) Permanent mandibular advancement device.

(Adapted from: Fetner M, Cascio CJ, Essick G. Nonverbal patient with autism spectrum disorder and obstructive sleep apnea: use of desensitization to acclimatize to a dental appliance. Pediatr Dent. 2014 Nov-Dec;36(7):499-501. PMID: 25514080.)

Outcome of Case

Thirty-three months after the initial appointment, the patient came in for a follow-up visit and his mother reported that the new appliance was working well.

A month later, the patient underwent a post-treatment sleep study. His BMI had remained largely stable over the three-and-a-half-year period between sleep studies. Based on the study results, the sleep physician reported that the sleep apnea was significantly improved with use of the oral appliance. There were only 6 obstructive, zero mixed, and 33 hypopneas. The AHI was 4.6 for sleep in the supine position. The oxygen saturation averaged 97.1 percent.

Teaching Points

This study is particularly illustrative, considering the patient was low-functioning and non-verbal and, thus, faced more significant challenges to treatment than the average ASD patient. Sleep problems are one of the most common complaints in autistic adults with prevalence rates of sleep problems reported as high as 79%. Important to note, his seizures were partial and under control for several years with no nocturnal seizures documented. In patients with nocturnal and generalized tonic-clonic seizures an oral device is not advisable.



This case report demonstrates that understanding the specific challenges inherent to ASD patients and the foresight and patience of providers in approaching these challenges, in collaboration with caregivers, can contribute to improved health outcomes.

The use of an oral appliance to ameliorate sleep apnea may have wide applicability to the ASD population, given the prevalence of sleep disturbances in ASD. It is important to note that caregiver involvement is critical for the success of this kind of behavioral approach. Incorporation of practice devices that were taken home with the patient and their regular use by the patient's mother in preparation for each stage of the protocol allowed the devices to become familiar and routine. Weight loss must be included in any treatment for OSA.

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Additional Reading

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