CASE REPORTS

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# Sleep Apnea in Fragile X Syndrome: Case Report

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This is the report of oral appliance therapy in an individual with fragile X syndrome, diagnosed with severe OSA who refused CPAP therapy.

**Keywords:** fragile X, oral appliance therapy, genetic disorders

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Fragile X syndrome (FXS) is a genetic disorder that occurs in approximately 1 in 4,000 males and 1 in 8,000 females.[1] It is an X-linked dominant disorder that is associated with syndromic intellectual disabilities. The disorder is associated with learning disabilities, attention deficit disorders, and autism spectrum disorders. Seizures occur in 5% to 15% of those with fragile X syndrome. Physical features common with the disorder include an elongated face, high arched palate, large or protruding ears, and macroorchidism in males after puberty.[2]

Subjects with fragile X are at an increased risk for OSA, and physicians should orient their evaluation with this in mind. Over 50% of those with fragile X suffer from OSA. They are found to have a reduction in REM sleep percentages that leads to intellectual disability and epilepsy.[3]

## REPORT OF CASE

A 47-year-old independent living male with FXS presented with OSA. He had been diagnosed by polysomnogram (PSG) with severe apnea: AHI 55.7, RDI 59.5, and lowest oxygen saturation of 83%. He was placed on CPAP therapy of 12 cm of water; he refused to use CPAP after the first night.

The patient lived in a group home where he had been noted to be a loud snorer and often was observed to stop breathing multiple times a night. His medical conditions included anxiety, depression, heartburn, hypertension, intellectual disability, and psychosis. His medications included divalproex, trazodone, Seroquel, Prilosec, levothyroxine, and hydrochlorothiazide. His caretaker reported that the patient was always tired and slept a lot during the day.

Oral exam revealed an enlarged scalloped tongue, type 3 Mallampati, and missing tonsils. He had a Class I occlusion with an anterior open bite with −2 mm overbite and +4 mm overjet with a very narrow palate and lower jaw. His dentition was in adequate condition with no sign of decay or periodontal disease. Bilateral crepitus of both joints on opening and closing with mild muscle tenderness was noted on examination. Very good range of motion was also noted on examination, with constant movement of his lower jaw laterally. There was very little wear on his teeth.

Before initiating treatment with an oral appliance (OA), we wanted to determine if the patient could tolerate wearing a device in his mouth on a consistent basis at night. We constructed an upper and lower sports mouth guard to wear for about one month. As a side note, we made the mouth guards the color of his favorite baseball team to engage and encourage the patient. After 1 month, the patient and caregiver returned and reported he was able to wear the mouth guards on a consistent basis with no side effects.

A Herbst-style OA was fabricated without elastics to allow the patient the freedom to move his jaw laterally. Extra strong bars had to be added to his OA as after 1 month of use as he had bent the original bars. The patient was initially started at 7 mm from his maximum protrusion. His OA was subsequently titrated 4 mm further to help with daytime sleepiness and snoring.

At subsequent follow-up appointments, his caregiver reported the patient was snoring significantly less and is less tired during the day. It was also noted the patients was less irritable and easier to communicate with on a consistent basis. The patient has not returned for a follow-up sleep study, as he adamantly refuses to undergo another sleep test.

Treatment of patients with FXS or other disabilities can be very rewarding but also frustrating at times. It is often necessary to alter the sequence and pace of routine OA protocol. All staff members should be educated to expect and accept the likely need for more frequent than usual follow-up visits to maximize treatment benefit.

## REFERENCES

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## SUBMISSION & CORRESPONDENCE INFORMATION

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## DISCLOSURE STATEMENT

Dr. Postol authors has indicated no financial conflicts of interest.