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SLEEP MEDICINE PEARLS

An Unusual Cause of Obstructive Sleep Apnea in a Man With Spinal Muscular Atrophy Type III

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A man with spinal muscular atrophy type III complained of snoring since age 18 years, shortly after he became wheelchair bound. He was not obese and allergic rhinitis was mild. His tonsils were not enlarged. Polysomnography (PSG) performed at age 18 years confirmed mild obstructive sleep apnea (OSA) but the patient opted for conservative management. A repeat PSG at age 20 years showed worsening OSA with apnea-hypopnea index of 23.5 events/h and oxygen saturation nadir of 76%. The patient began nocturnal continuous positive airway pressure at age 21 years with satisfactory titration at 5 cmH_2O.

The patient's OSA was initially attributed to the onset of respiratory muscle weakness because there was a simultaneous drop in his lung function parameters after he became non-ambulatory (**Table 1**). There was some initial improvement with the use of continuous positive airway pressure, but overnight oximetry repeated 1 year later showed deterioration with more desaturations and paired morning arterial blood gas revealed elevated PCO_2 to 6.4 kPa, suggestive of nocturnal hypoventilation. With the patient's history of neuromuscular disease, he was advised to switch to bilevel positive airway pressure. Again, there was initial improvement with low ventilator settings (S/T mode, IPAP 8, EPAP 4, rate 9/min, inspiratory time 1 second, rise time 2) but repeated monitoring showed deterioration after a few months. His OSA symptoms (snoring, mouth breathing, daytime somnolence) also continued to progress even though lung function parameters were relatively maintained during that period. However, the patient could not tolerate escalation of pressure support with bloating sensation; therefore, the titration was not optimal and his bilevel positive airway pressure compliance was poor. At clinic follow-up at age 23 years, the patient reported a globus sensation at the back of his tongue for a few months accompanied by dysarthria. Examination revealed that his tongue was mildly deviated to the right, although there was no obvious palpable mass. Magnetic resonance imaging (MRI) was performed (**Figure 1**).

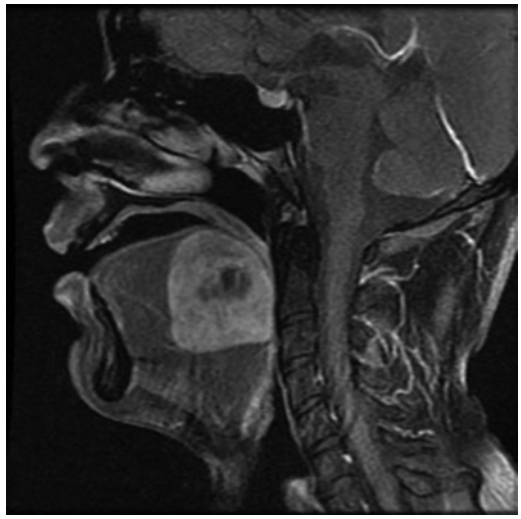
QUESTION: What was the cause of the patient's OSA progression?

Table 1—Serial respiratory function and overnight sleep parameters.

Status		Ambulatory	Non-ambulatory							
Age (years)		14	18	19	20	21–22	22–23		Surgery	24
Symptomatology		Nil	Snoring		Increase in snoring, daytime somnolence		Insidious onset of tongue mass and dysarthria			Snoring resolved
Body weight (kg)		39.2	43.0	43.6	44.0	44.5	45.0			48
Erect FEV_1 (liter)		2.18	1.92	1.53	1.52	1.64	1.72			1.62
Erect FVC (liter)		2.25	1.94	1.61	1.59	1.8	1.82			1.80
PCF (L/min)		230	194	188	195	195	222			232
MIP/MEP (cmH_2O)		60/60	65/44	46/39	–	39/38	39/36			37/35
Ventilatory support		Nil (conservative)				CPAP	Switched to BPAP		Off BPAP postop	Nil
PSG	Obstructive AHI (events/h)	–	15	–	23.5	–	–			5.2
	Nadir SaO_2 (%)	–	87	–	76	–	–			85
Overnight oximetry	No. of desaturation charted per night (defined as SaO_2 drop > 4% compared to baseline SaO_2)	9	55	65	–	3	28	15	74	14
	Nadir SaO_2 (%)	91	89	87	–	93	82	92	87	89
PCO_2 (kPa)		–	5.0	5.4	–	5.4	6.4	5.6	6.6	5.3

AHI = apnea-hypopnea index, BPAP = bilevel positive airway pressure, CPAP = continuous positive airway pressure, FEV_1 = forced expiratory volume in 1 second, FVC = forced vital capacity, MEP = maximum expiratory pressure, MIP = maximum inspiratory pressure, PCF = peak cough flow, PCO_2 = partial pressure of carbon dioxide, PSG = polysomnography.

Figure 1—MRI T1 sagittal view (post-contrast).



MRI T1 sagittal view (post-contrast) showed an oval 5 cm × 4 cm × 4 cm tumor in the posterior third of the tongue. MRI = magnetic resonance imaging.

ANSWER: Posterior lingual tumor causing upper airway obstruction.

MRI showed a tumor at the posterior right genioglossus muscle (**Figure 1**). The lingual tumor was excised and pathology result confirmed the presence of schwannoma. Snoring resolved and a repeat PSG 7 months after the surgical procedure showed reduction of apnea-hypopnea index to 5.2 events/h with no significant desaturations. It was difficult to ascertain the exact onset of this slow-growing tumor. Yet, excision resulted in significant improvement of the patient's OSA and suggested that the tumor could be the key culprit.

DISCUSSION

Patients with neuromuscular disorders are vulnerable to sleep-disordered breathing, because of a combination of alveolar hypoventilation resulting from respiratory muscle weakness and increased upper airway resistance secondary to pharyngeal dilator muscle atonia.[1,2] In our patient, OSA developed during the time of transition to non-ambulatory state but there was unexpected progression of OSA in the absence of ongoing respiratory muscle weakness. A second pathology, lingual schwannoma, was found to be the main culprit for the patient's OSA.

Schwannoma is a rare, benign tumor composed of Schwann cells. Only 1% of these tumors occur in the oral cavity, with the tongue being the most common site. Presenting symptoms of lingual schwannoma vary and include nodules, dysphagia, bleeding, dysphonia, and rarely, sleep apnea. Malignant transformation is rare and surgical excision is the mainstay of treatment.[3,4] Some patients may experience OSA for years before some slow-growing tumors in the head and neck regions are diagnosed.[5]

SLEEP MEDICINE PEARLS

1. Patients with neuromuscular weakness are prone to sleep-disordered breathing but the clinician should be alerted to other possible cause(s) when the deterioration of OSA is out of proportion to respiratory function.
2. Head and neck tumor can be a rare cause of OSA and should be considered in the differential diagnosis when evaluating a patient with refractory symptoms. A comprehensive head and neck evaluation, assisted by imaging or endoscopy, is required for early diagnosis and subsequent intervention.

CITATION

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DISCLOSURE STATEMENT

All authors have seen and approved the manuscript. All authors report no conflicts of interest. Patient consent was obtained for publication of this case.