TITLE:
Palatal and diaphragmatic myoclonus with sleep disordered breathing

Authors:
Ramesh Donepudi, MD\textsuperscript{1}, Karin G Johnson, MD\textsuperscript{1}, Asha Singh, MD\textsuperscript{1}, Jean K Matheson, MD\textsuperscript{1}

Institution:
Beth Israel Deaconess Medical Center\textsuperscript{1}, Boston, MA

Disclosures: None

Corresponding Author:
Ramesh Donepudi, MD
Beth Israel Deaconess Medical Center
Harvard Medical School
330 Brookline Ave
Boston, MA 02215

Phone:617-667-3237
Fax: 617-975-5506
Email: rdonepud@bidmc.harvard.edu
ABSTRACT

We describe a case of an 83-year-old male presenting with excessive daytime somnolence, ataxia, palatal and diaphragmatic myoclonus and polysomnographic features of periodic breathing, who benefited from treatment with adaptive servo ventilation. Diaphragmatic myoclonus has been reported in the setting of palatal myoclonus, a disorder associated with degenerative hypertrophy of the inferior olive of the medulla. We propose a disruption of respiratory control pathways leading to sleep disordered breathing in association with this uncommon clinical entity.

Key Words: Palatal myoclonus, Respiratory myoclonus, Periodic breathing
INTRODUCTION

Antony van Leeuwenhoek first described diaphragmatic myoclonus in 1723 after developing the symptom complex\(^1\). Prior cases of shortness of breath and diaphragmatic clicking have been described, but there has been no reported case of palatal and diaphragmatic myoclonus associated with sleep-disordered breathing (SDB). Recognizing this association is important since SDB is treatable with positive airway pressure.

CASE REPORT

An 83 year old, Caucasian gentleman presented with three years of progressive hypophonia, gait and balance difficulty, and palatal myoclonus. The patient complained of un-refreshed sleep, excessive daytime sleepiness with an Epworth Sleepiness score of ten. His wife noted loud snoring, periods of apneas and intermittent bursts of rapid breathing with pulsations of his epigastrium. He denied vertigo, or swallowing problems but admitted having diplopia and hearing loss in his left ear prior to presentation. His exam showed bilateral finger nose dysmetria, wide base gait, impaired abduction of left eye and continuous palatal myoclonus (1-2Hz). Cardiovascular examination was normal with no signs of congestive heart failure. A diagnostic polysomnogram showed severe SDB with an apnea-hypopnea index of 72.9 events /hr (combination of central and obstructive apneas and hypopneas as described by loss of nasal pressure signal of 50% associated with 4% reduction in oxygen saturation). There was also evidence of periodic breathing pattern and bursts of fragmentary myoclonus of the thoraco-abdominal signals suggestive of underlying diaphragmatic myoclonus(Figure 1)
MRI of brain revealed generalized atrophy, bilateral symmetric olivary hypertrophy and scattered high T2 signals suggestive of small vessel disease. Serum B12, red cell folate, ceruloplasmin and BNP levels were normal. Lung function tests revealed mild restrictive physiology with normal diffusion capacity. Echocardiogram was normal.

He was started on CPAP at 15 cm of water pressure at another institution, with inadequate response. He was re-titrated with adaptive servoventilation (ASV) in our sleep laboratory, which led to stabilization of his breathing pattern, eliminating central apneas and improvement of daytime sleepiness.

DISCUSSION
Symptomatic palatal myoclonus is characterized by rhythmic 1-Hz movement of the posterior palate (levator veli palatini muscle), innervated by cranial nerves IX and X (nucleus ambiguous). Most patients with palatal myoclonus have structural abnormalities involving the connections between the dentate, red and inferior olivary nuclei (Mollaret's triangle) leading to denervation supersensitivity of the inferior olivary nucleus and olivary pseudohypertrophy. It is sometimes associated with ataxia, tremor, diplopia and involvement of other neck muscles.

Respiratory myoclonus results from spread of the contraction to the diaphragm. The diaphragm is dually innervated by the motor cortex through the corticospinal tract and the medullary respiratory centers, including the nucleus ambiguous, retroambigualis, and para-ambiguus of the ventral respiratory group and the nucleus of the solitary tract of the dorsal respiratory group. Differing supra-spinal mechanisms have been speculated to be
responsible for normal diaphragmatic function in comparison to those involved in a pathological state. Degeneration of the nucleus ambiguus and dorsolateral reticular formation have been proposed to cause palatal myoclonus.

Central sleep apnea with periodic breathing pattern has been reported in patients who had brainstem infarcts. Elevated hypercapnic ventilatory responses leading to hypocapnia and respiratory control instability are believed to be particularly important. Arousal and accompanying hyperventilation likely play an important role in triggering hypocapnia, resulting in further destabilization if breathing. In our case all the above factors viz. altered apnea threshold, arousal threshold and ventilatory response to arousal could be playing a role in emergence of periodic breathing. Alternatively, uninhibited olivary bursting into deep cerebellar nuclei could be triggering diaphragmatic myoclonus and resulting periodic breathing. It has been shown in animal models that vestibular nuclei and fastigial nuclei play an important role in respiratory control in hypercapnic and hypoxic conditions.

Recognizing the association between SDB and palatal and diaphragmatic myoclonus is important, as SDB is now potentially treatable with various modalities to improve patient morbidity and mortality. Our patient improved with ASV, which is a form of dynamic bi-level positive pressure that targets minute ventilation. Compared to delivering a continuous pressure with conventional CPAP, ASV delivers a varying amount of tidal volume dependent on an averaged minute ventilation, thereby improving treatment of central apneas.
REFERENCES


A 2 minute window of PSG showing fragmentary myoclonus (shaded area) and periodic breathing pattern. Channels in order: Hypnogram, C3A2- parietal EEG, O1A2-occipital EEG, THO- thoracic movements, ABD-abdominal movements, SpO2-oxygen saturation, NAF-nasal airflow pressure, Stage-sleep stage