Puzzling Dyspnea Caused by Respiratory Muscle Weakness

Adriana J. Pavletic and Oleh Hnatiuk

Dyspnea is common in advanced stages of neuromuscular disorders, but it is infrequently the presenting symptom. However, dyspnea is a frequent complaint in a primary care setting but is rarely caused by a respiratory muscle weakness. Consequently, the diagnosis of respiratory muscle weakness often is delayed. First symptoms may occur when respiratory muscles are under increased load, such as when standing in the water higher than the chest, swimming, or in the supine position. We describe a patient in whom dyspnea was the first symptom of amyotrophic lateral sclerosis to remind clinicians of clinical features of respiratory muscle weakness and to help avoid the delay in diagnosis. (J Am Board Fam Med 2012;25:396–397.)

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Although dyspnea is a frequent complaint in a primary care setting, it is rarely caused by a respiratory muscle weakness. Consequently, physicians often are not familiar with the clinical presentation of respiratory muscle dysfunction and it may be wrongly attributed to other causes. To remind physicians about this potentially puzzling condition, we describe a patient in whom dyspnea was the first symptom of amyotrophic lateral sclerosis (ALS).

A 79-year-old, white, nonsmoking, physically fit man complained of progressive shortness of breath for 1 year. He had well-controlled hypertension, type 2 diabetes mellitus, and dyslipidemia, for which he took lisinopril, metformin, and simvasta-
pressure was +68 (normal, +140 to +190). His daytime fatigue temporarily improved with the nighttime use of bilevel positive airway pressure, but his illness relentlessly progressed and he died of respiratory failure 3 years after the onset of symptoms.

Respiratory muscle dysfunction is common among patients with many neuromuscular disorders. It is rarely a presenting symptom and therefore often goes unrecognized. Clinical presentation can be chronic progressive (eg, ALS), acute (eg, Guillain–Barré syndrome), or chronic relapsing (eg, myasthenia gravis, multiple sclerosis). Clinical course, prognosis, and treatment are variable and depend on the underlying disease.

Patients with motor neuron disease typically complain of the inability to take a deep breath, which may be confused with anxiety. However, their symptoms are worse in a supine position, which is not a feature of psychogenic dyspnea. Our patient’s initial symptom of dyspnea while immersed in water likely was caused by diaphragmatic dysfunction.

In one study involving patients with multiple sclerosis, a clinical assessment index was found to better predict respiratory muscle weakness than spirometry. This index comprised of patients’ reports of a weakened cough and difficulty clearing pulmonary secretions, weakened cough as rated by the examiner, and the counting test (value reached when the patient counts aloud on a single expiration after maximum inspiratory effort; normal ≥30). Pulmonary function tests in patients with advanced respiratory muscle weakness have a restrictive pattern (reduced FVC and total lung capacity, normal FEV₁/FVC) and reduced maximal inspiratory pressure and maximal expiratory pressure. Vital capacity measured in the supine position is usually lower than in the upright position.

Respiratory muscle weakness should be considered in patients whose dyspnea cannot be explained by pulmonary disease, cardiac disease, anemia, or deconditioning. First symptoms may occur when respiratory muscles are under increased load, such as when standing in deep water, swimming, or in the supine position.

References