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Thinking Outside the Cardiac Box: Anchoring in an Elderly Patient with Multiple Visits for Orthopnea

## Case information:

A 77-year-old male presented four times over four months for evaluation of progressive shortness of breath worse with lying down and associated with fatigue. His pmhx included HTN and prostate cancer. During his first visit, he was discharged from the ED with dyspnea of unknown etiology. On his next visit, he was admitted to the hospital where cardiac workup was unrevealing. On the third visit, the patient was discharged from the ED with a diagnosis of pneumonia, but returned six hours later for worsening cough and dyspnea. Oxygen saturation by pulse-ox during this presentation revealed mild hypoxia of 90% on room air while seated upright and 79% while supine. The patient was admitted to the hospital where workup was negative for pneumonia, COPD, heart failure, pulmonary shunting, and dysphagia. Detailed physical examination revealed fasciculations and weakness of the upper extremities, and hyperreflexia in the upper and lower extremities. Further discussion revealed four months of progressive weakness and several years of worsening tremors. Pulmonary function testing revealed reduced inspiratory strength and electromyography confirmed the diagnosis of amyotrophic lateral sclerosis (ALS).

## Discussion:

Dyspnea and fatigue are frequent complaints in outpatient settings. Though often due to cardiac dysfunction, the differential diagnosis for true orthopnea is limited and includes neuromuscular disease. This case illustrates the importance of maintaining a broad differential and considering a neurologic etiology for dyspnea. Although ALS has no cure to date, misdiagnosis can lead to delayed treatment, over-testing, reduced quality of life, and emotional distress for patients and their families.