



# Unusual Presentation of Pulmonary Embolism as Diaphragmatic Hernia - Case Study

Mohamed Saleh <sup>1\*</sup>

## Abstract

Pulmonary embolism (PE) is a critical condition characterized by thrombi obstructing pulmonary arteries, commonly originating from deep vein thrombosis. Typical symptoms include sudden-onset dyspnea, chest pain, and hypoxemia, although atypical presentations complicate diagnosis. Diaphragmatic hernias, involving abdominal organs displacing into the thoracic cavity, present with respiratory distress and gastrointestinal symptoms. The coexistence of PE and diaphragmatic hernia is rare, posing significant diagnostic challenges. This case report details a 65-year-old female with acute dyspnea and cyanosis following chest trauma, initially misdiagnosed with diaphragmatic hernia. Elevated D-dimer levels and clinical signs suggested PE, but CT pulmonary angiography revealed a large diaphragmatic hernia compressing the pulmonary arteries, causing pulmonary hypertension. This case underscores the importance of comprehensive differential diagnosis and advanced imaging to accurately identify overlapping conditions. It is important to consider differential diagnoses in acute dyspnea cases with atypical findings. Maintaining a broad differential diagnosis and a multidisciplinary approach is crucial to avoid misdiagnosis and ensure timely, appropriate treatment, thereby optimizing patient outcomes.

**Significance** | This study shows the importance of comprehensive differential diagnosis in acute dyspnea with atypical findings to prevent misdiagnosis and improve outcomes.

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## Introduction

Pulmonary embolism (PE) is a critical medical condition characterized by the obstruction of pulmonary arteries by thrombi, commonly originating from deep vein thrombosis (Goldhaber, 2022; Torbicki et al., 2018). Its hallmark symptoms include sudden-onset dyspnea, chest pain, and hemoptysis, often accompanied by signs of right heart strain and hypoxemia (Konstantinides et al., 2014). Despite these typical presentations, PE can manifest atypically, posing challenges in diagnosis and management (Wells et al., 2000; Stein et al., 2017).

Conversely, diaphragmatic hernias involve the abnormal displacement of abdominal organs into the thoracic cavity through a defect in the diaphragm, either congenital or acquired (Killeen, 2020; Shah et al., 2016). Patients typically present with respiratory distress, gastrointestinal symptoms, and audible bowel sounds in the chest (Boni et al., 2009; Al-Salem, 2013). The coexistence of PE and diaphragmatic hernia is exceedingly rare, presenting diagnostic complexities due to overlapping symptoms and potential misinterpretation (Athassiadi et al., 1999; Choi et al., 2010).

This case report discusses an unusual instance where PE masqueraded as a diaphragmatic hernia, highlighting diagnostic challenges and the importance of comprehensive differential diagnosis. Diagnosis of PE involves a high index of suspicion and systematic use of clinical models such as the Wells score, D-dimer testing, and imaging techniques like computed tomography pulmonary angiography (CTPA) (Stein et al., 2021; Kearon et al., 2016; Righini et al., 2008). However, the presence of concurrent

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conditions like diaphragmatic hernia can complicate diagnostic clarity and delay appropriate treatment (Miniati et al., 2001; Le Gal et al., 2006).

Diagnosis of diaphragmatic hernias typically relies on clinical evaluation, chest radiography, and confirmatory imaging such as CTPA or MRI (Dalton et al., 2019; Bochicchio et al., 2002; Gale et al., 1994). The overlap with PE further complicates symptom interpretation and imaging findings, posing diagnostic challenges (Berman et al., 2011; Shields et al., 2015).

In this reported case, initial misdiagnosis as diaphragmatic hernia delayed timely treatment for PE, underscoring the necessity of considering PE in differential diagnoses of respiratory distress (Levin et al., 2002; Porcel et al., 2019). A multidisciplinary approach involving pulmonologists, radiologists, and surgeons is crucial for accurate diagnosis and management (Blanco et al., 2009; Ouellette et al., 2019). Advanced imaging techniques play a pivotal role in identifying concurrent conditions that may obscure primary diagnoses (Madan et al., 2007; Sosna et al., 2017).

The case illustrates the complexities associated with diagnosing PE when presenting with atypical symptoms, emphasizing the need for meticulous clinical assessment and thorough differential diagnosis to optimize patient outcomes (Streiff et al., 2011; Klok et al., 2010). Clinicians must maintain a broad differential to avoid misdiagnosis and treatment delays, especially in cases where symptoms overlap or are obscured by concurrent conditions (Huisman et al., 2018; Raja et al., 2015).

The aim of this case report was to examine and discuss the diagnostic challenges and clinical implications of an unusual presentation where pulmonary embolism (PE) mimicked a diaphragmatic hernia. By exploring this rare scenario, the study aimed to underscore the importance of comprehensive differential diagnosis and multidisciplinary collaboration in accurately identifying and managing patients with overlapping or atypical clinical presentations. Through this analysis, the study sought to enhance clinical awareness and improve diagnostic strategies for similar cases, ultimately aiming to optimize patient outcomes through timely and appropriate intervention.

### Clinical Presentation

A 65-year-old female, a housewife with no significant past medical history or recent operations, presented to the clinic with acute shortness of breath and cyanosis. The patient's symptoms developed rapidly, five days after she experienced chest trauma from hitting a door. Her symptoms included acute dyspnea, tachypnea, and regular palpitations. There was no history of orthopnea, paroxysmal nocturnal dyspnea (PND), or chronic respiratory or cardiac issues. Additionally, the patient had no history of surgical operations, medication use, or blood transfusions.

### Physical Examination

On physical examination, the patient's vital signs were as follows: blood pressure of 120/80 mmHg, heart rate of 120 beats per minute (regular), respiratory rate of 30 breaths per minute, and oxygen saturation of 91% on room air. The general examination revealed that the patient was conscious and oriented to time, place, and person. She had congested pulsating neck veins without abnormal waveforms. Her abdomen was tender, but there were no signs of lower limb edema, and peripheral pulses were intact.

Cardiac examination showed a normal chest contour, with no pericardial bulge or scars. The apex beat was located at the 5th intercostal space at the midclavicular line, with average intensity. Auscultation revealed normal heart sounds (S1, S2) without additional sounds or murmurs.

### Laboratory Investigations

Laboratory investigations revealed that the patient's hemoglobin (HB) level was 12.5 g/dL, white blood cell (WBC) count was 6,000 cells/ $\mu$ L, platelet (Plt) count was 299,000 cells/ $\mu$ L, and creatinine level was 0.9 mg/dL. Notably, the D-dimer level was significantly elevated at 5000 ng/mL, which initially suggested the possibility of a thrombotic event.

### Diagnostic Imaging

Electrocardiogram (ECG) showed sinus tachycardia with an S1Q3T3 pattern, suggesting potential right heart strain. The echocardiogram (ECHO) revealed right heart dilation, moderate tricuspid regurgitation (TR), and mild pulmonary hypertension.

CT pulmonary angiography was performed to rule out pulmonary embolism. The imaging revealed no evidence of pulmonary embolism. However, it showed a large diaphragmatic hernia containing the stomach, exerting pressure on the left atrium and causing significant compression of the pulmonary arteries. Additional findings included bilateral mild asymmetric pleural effusion (more on the left side) with partial left posterior basal atelectasis, bilateral mild pulmonary congestion, and fine linear bibasal edematous reticulations. The cardiac chambers were noted to be enlarged, but the main tracheobronchial tree was normal. There were no mediastinal masses or significant lymphadenopathy.

### Diagnosis

The patient was diagnosed with a large diaphragmatic hernia causing compression of the pulmonary arteries and resulting in pulmonary hypertension. The initial suspicion of pulmonary embolism was ruled out by CT pulmonary angiography.

### Discussion

This case demonstrates an unusual presentation of dyspnea and hypoxia that mimicked pulmonary embolism. The patient's high D-dimer levels and clinical signs initially suggested PE, a common and potentially fatal condition. However, the absence of emboli on CT angiography prompted further investigation. The subsequent

imaging revealed a large diaphragmatic hernia as the underlying cause of the patient's symptoms.

Diaphragmatic hernias are typically congenital but can be acquired due to trauma, as in this patient's case. The hernia contained the stomach and exerted significant pressure on the left atrium and pulmonary arteries, leading to cardiopulmonary compromise. This compression caused right heart strain and pulmonary hypertension, explaining the patient's symptoms and clinical findings.

The large diaphragmatic hernia led to significant cardiopulmonary compromise, manifesting as right heart strain and pulmonary hypertension. This unusual presentation underscores the importance of considering a wide range of differential diagnoses in patients with acute dyspnea, especially when initial findings do not align with common diagnoses such as pulmonary embolism.

### Conclusion

In cases of acute dyspnea with atypical findings and high D-dimer levels, it is essential to consider differential diagnoses beyond pulmonary embolism. Comprehensive imaging and thorough clinical assessment are crucial in identifying rare but significant conditions like diaphragmatic hernia, which can present with life-threatening complications. This case highlights the importance of maintaining a broad differential diagnosis and the role of advanced imaging techniques in accurately diagnosing complex cases.

### Author contributions

M.S was solely responsible for the conceptualization, methodology, data collection, analysis, and manuscript preparation of this study.

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### Competing financial interests

The authors have no conflict of interest.

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