UNDER PEER REVIEW IN IJARSpontaneous Pneumomediastinum Revealing Diffuse Cystic Lung Disease in a Young Woman: A Case Suggestive of Lymphangioleiomyomatosis

Abstract:
Spontaneous pneumomediastinum is a rare clinical entity characterized by the presence of air in the mediastinum without preceding trauma or invasive procedures. It predominantly affects young adults and usually has a benign outcome. We report the case of a 22-year-old woman presenting with sudden retrosternal chest pain and dyspnea, whose chest CT revealed pneumomediastinum associated with cervical and thoracic subcutaneous emphysema. Conservative treatment led to clinical improvement, but follow-up imaging uncovered diffuse bilateral thin-walled pulmonary cysts suggestive of lymphangioleiomyomatosis (LAM). Pulmonary function tests showed severe ventilatory impairment, and the patient was referred for specialized care. This case highlights that spontaneous pneumomediastinum may be the first manifestation of underlying cystic lung disease, particularly LAM in young women. Early recognition through imaging and clinical evaluation is crucial for diagnosis and management.

Keywords: pneumomediastinum, lymphangioleiomyomatosis (LAM), diffuse cystic lung disease, subcutaneous emphysema, respiratory function impairment

Introduction:

Spontaneous pneumomediastinum is a rare condition characterized by the presence of air in the mediastinum without any history of trauma or invasive procedures. Its incidence is estimated to range from 1 in 7,000 to 1 in 45,000 hospitalizations [1]. The condition primarily affects young adults and generally follows a benign course. Patients most commonly present with sudden chest pain, shortness of breath, and occasionally subcutaneous emphysema in the cervical or thoracic regions [2].

The underlying mechanism is often explained by the Macklin effect, in which a sudden rise in intra-alveolar pressure causes alveolar rupture, allowing air to track along the bronchovascular sheaths toward the mediastinum [3]. While Spontaneous pneumomediastinum is idiopathic in most cases and usually resolves with conservative measures, it can sometimes reveal underlying pulmonary disorders, particularly diffuse interstitial or cystic lung diseases [4].

Case Presentation:

A 22-year-old woman, a nonsmoker with no significant past medical history, presented to the emergency department with sudden retrosternal chest pain accompanied by dyspnea.

On initial clinical examination, the patient was hemodynamically stable with an oxygen saturation of 94% on room air. Physical examination revealed cervical and thoracic subcutaneous emphysema. Pulmonary auscultation revealed no significant abnormalities.

An emergency chest CT angiogram revealed pneumomediastinum associated with diffuse cervical and thoracic subcutaneous emphysema, with no signs of pulmonary embolism or pleural effusion (Figure 1, Figure 2).

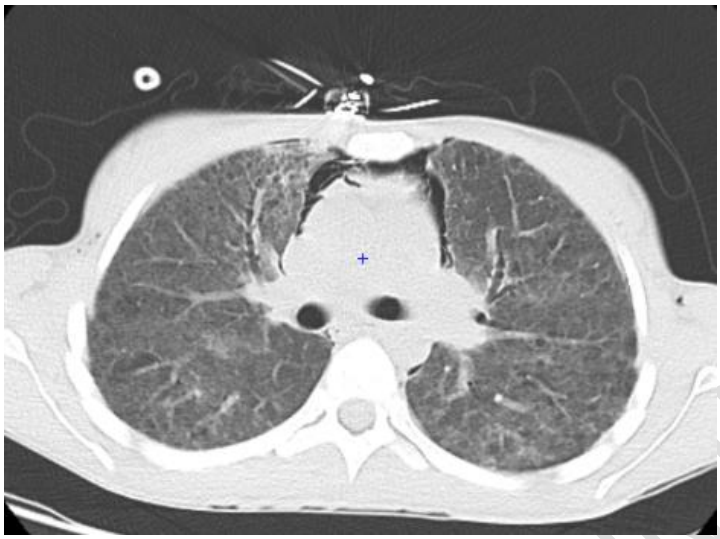

Figure 1. Axial chest CT scan demonstrating pneumomediastinum

Figure 2. Axial chest CT scan demonstrating pneumomediastinum associated with subcutaneous emphysema of the chest wall

To rule out any secondary causes, a bronchoscopy and an upper gastrointestinal endoscopy were performed, which revealed no tracheobronchial fistula or esophageal perforation. (Figure 3)

Figure 3. Bronchoscopic view showing inflamed bronchial mucosa without any evidence of tracheobronchial breach

The patient received conservative management consisting of rest and clinical monitoring. The course of the illness was favorable, with a gradual improvement in symptoms.

A follow-up chest CT scan revealed that the pneumomediastinum and subcutaneous emphysema had completely resolved. However, this scan revealed a diffuse bilateral pulmonary microcystic syndrome, characterized by multiple spherical cysts of relatively uniform size distributed throughout the lung parenchyma. This radiological finding was highly suggestive of lymphangioleiomyomatosis.(Figure 4)

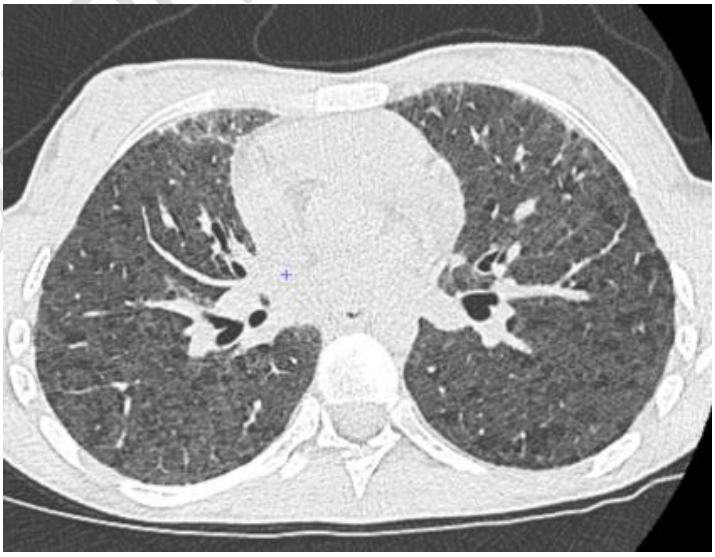

Figure 4: Axial high-resolution chest CT scan showing multiple bilateral thin-walled pulmonary cysts diffusely distributed throughout the lung parenchyma, suggestive of lymphangioleiomyomatosis

Pulmonary function tests revealed severe ventilatory impairment, with an FEV_1 at 27% of the predicted value and a vital capacity at 23%. Alveolar-capillary diffusion could not be measured.

Arterial blood gas analysis at rest showed a PaO_2 of 76 mmHg with an oxygen saturation of 94.8%. During the six-minute walk test, the patient walked 280 meters with a drop in oxygen saturation from 94% to 87%, accompanied by significant dyspnea. A contrast-enhanced abdominal CT scan did not reveal a renal angiomyolipoma.

Based on these clinical, functional, and radiological findings, a diagnosis of diffuse cystic lung disease, likely lymphangioleiomyomatosis, was strongly suspected, and the patient was referred to a specialized center for diagnostic confirmation and specialized care.

Discussion:

Pathophysiology

The development of Spontaneous pneumomediastinum is typically linked to abrupt increases in intrathoracic pressure. This can occur during episodes of severe coughing, vomiting, asthma attacks, Valsalva maneuvers, or intense physical exertion [2]. In many cases, however, no clear precipitating event is identified. Air from ruptured alveoli dissects along the bronchovascular pathways and accumulates in the mediastinum, as described by Macklin [3].

Diagnosis

Diagnosis relies primarily on imaging studies. Although chest X-rays may occasionally detect mediastinal air, computed tomography (CT) is the preferred method for confirming the diagnosis and for identifying any underlying causes [5]. CT imaging also allows assessment of subcutaneous emphysema and exclusion of complications such as pneumothorax or esophageal perforation.

Clinical Course and Management

Spontaneous pneumomediastinum generally has a favorable prognosis. Most patients recover with conservative treatment, which includes rest, oxygen supplementation, and careful monitoring [6]. Follow-up imaging is sometimes necessary to identify previously unrecognized pulmonary abnormalities.

Spontaneous pneumomediastinum in Cystic Lung Diseases

Diffuse cystic lung diseases, such as lymphangioleiomyomatosis (LAM), pulmonary Langerhans cell histiocytosis, and Birt-Hogg-Dubé syndrome, are characterized by the presence of multiple cysts within the lung parenchyma [7]. Among these, LAM predominantly affects women of childbearing age and involves proliferation of abnormal smooth muscle cells, leading to progressive lung destruction [3].

High-resolution CT is key for diagnosis, revealing numerous thin-walled cysts throughout both lungs. In some cases, serum VEGF-D can serve as a biomarker, potentially avoiding the need for lung biopsy [8]. Although pneumothorax is a well-known complication of LAM, pneumomediastinum remains uncommon but can be explained by the fragility of cystic lung tissue [9].

Conclusion:

Spontaneous pneumomediastinum is an uncommon clinical condition that generally follows a benign course and responds well to conservative management. Nevertheless, it may occasionally represent the initial manifestation of an underlying pulmonary disorder.

In young women, the incidental discovery of diffuse pulmonary cysts on imaging should prompt consideration of lymphangioleiomyomatosis. Careful analysis of chest CT findings and appropriate specialized evaluation are essential to establish the diagnosis and ensure adequate long-term management.

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