

THE CLINICAL PICTURE

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The Clinical Picture

Recurrent spontaneous pneumothorax



FIGURE 1

A 26-YEAR-OLD WOMAN presents with the sudden onset of left chest pain and dyspnea while at rest. She has never smoked. Two months ago, she developed a right-sided pneumothorax, which was treated with a tube thoracostomy. At that time, she received a diagnosis of tuberous sclerosis complex.

She has no other medical problems, has no history of trauma, and has tested negative for human immunodeficiency virus (HIV).

On physical examination, her breath sounds in the left hemithorax are decreased. She also has many nodular lesions on her face (FIGURE 1), a hypopigmented lesion on her right thigh (FIGURE 2), and a fleshy plaque on her lower back (FIGURE 3).

Radiography (FIGURE 4) and computed tomography (CT) of the chest (FIGURE 5) reveal a left-sided pneumothorax, a small right-sided pneumothorax, and diffuse, bilateral, thin-walled cysts in the lung.



FIGURE 2



FIGURE 3

Q: Which is the most likely diagnosis?

- Pulmonary Langerhans cell histiocytosis
- Cystic fibrosis
- Pneumocystis jirovecii* pneumonia
- Alpha-1 antitrypsin deficiency
- Tuberous sclerosis complex with lymphangiomyomatosis

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FIGURE 4. Left-sided pneumothorax (arrow) on chest radiography at presentation.

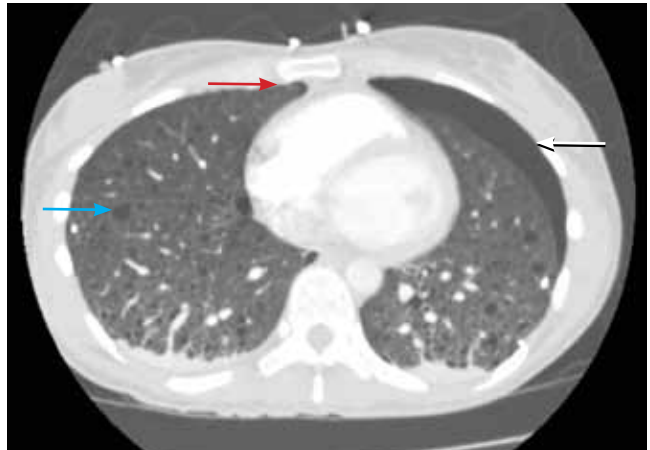


FIGURE 5. Computed tomography shows a left-sided pneumothorax (white arrow), a small right-sided pneumothorax (red arrow), and diffuse bilateral thin-walled cysts (blue arrow) in the right lung.

A: The correct diagnosis is tuberous sclerosis complex with lymphangiomyomatosis. The lymphangiomyomatosis was suggested by the CT findings, by the recurrence of pneumothorax, and, later, by biopsy results. Lymphangiomyomatosis occurs in about 30% of women with the tuberous sclerosis complex.¹ However, 10% to 15% of women with lymphangiomyomatosis do not have tuberous sclerosis complex,² in which case the condition is called sporadic lymphangiomyomatosis.

Tuberous sclerosis complex can involve the nerves (seizures, brain tumors), the lungs (lymphangiomyomatosis, causing pneumothorax or chylothorax), and the skin; skin lesions include facial angiofibromas (FIGURE 1), ash-leaf spot (FIGURE 2), and shagreen patch (FIGURE 3). It is also associated with abdominal involvement (lymphangiomyomas, renal angiomyolipomas).³

Lymphangiomyomatosis usually presents as spontaneous pneumothorax in women of childbearing age. After initial stabilization of pneumothorax with simple aspiration or thoracostomy, the patient should undergo ipsilateral chemical or surgical pleurodesis, as the risk of recurrent pneumothorax is greater than 70%.⁴ Single or bilateral lung transplantation has been accepted as therapy for end-stage pulmonary lymphangiomyomatosis, characterized by recurrent pneu-

mothoraces and chylous pleural fluid collections causing respiratory failure (marked dyspnea, hypoxemia, and reductions in forced expiratory volume in the first second of expiration and in diffusing capacity for carbon monoxide). Recurrence of lymphangiomyomatosis in the allograft lung is rare.⁵ Hormone therapies such as intramuscular progesterone, oral progestins, or gonadotropin-releasing hormone agonists have been used for lymphangiomyomatosis (not pneumothorax), but they are no longer recommended.

Pulmonary Langerhans cell histiocytosis, cystic fibrosis, *Pneumocystis jirovecii* pneumonia, and alpha-1 antitrypsin deficiency have all been associated with spontaneous pneumothorax.

Pulmonary Langerhans cell histiocytosis can affect multiple systems, including lung, skin, bone, and the pituitary gland. More than 90% of patients have a history of smoking.⁶ Chest radiography reveals a reticulonodular pattern with involvement of the middle and upper lobes. Later, the nodules tend to cavitate and form contiguous cysts that may mimic lymphangiomyomatosis on a high-resolution chest CT.

Cystic fibrosis is most often diagnosed before the age of 3.⁷ In adults, it can present as sinus and pulmonary disease (chronic cough with sputum production, chronic sinusitis with nasal polyposis, radiographic evidence of bronchiectasis and, less commonly, pneumothorax); as a gastrointestinal tract and nutritional abnormality (pancreatic insufficiency, distal intestinal obstruction, focal biliary cirrhosis); and as male infertility.

***Pneumocystis jirovecii* pneumonia** occurs mainly in patients on chronic immunosuppressive drugs or with immune deficiency due to HIV infection. Typical radio-

graphic features are bilateral perihilar interstitial infiltrates that become increasingly homogeneous and diffuse as the disease progresses. Less common findings include solitary or multiple nodules, upper-lobe infiltrates in patients receiving aerosolized pentamidine (NebuPent), pneumatoceles, and pneumothorax.⁸

Alpha-1 antitrypsin is an inhibitor of neutrophil elastase. Deficiency is associated with severe, early-onset panacinar emphysema with a basilar predominance, with chronic liver disease including cirrhosis, and less commonly with panniculitis and vasculitis associated with antineutrophil cytoplasmic antibody.⁹ Coalescence of panacinar emphysema leads to the formation of bullae and is important in the development of spontaneous pneumothorax.¹⁰

The patient underwent bilateral talc pleurodesis. Lung biopsy at the same time confirmed lymphangioleiomyomatosis. One month later, the right pneumothorax recurred, and she underwent pleurodesis in the right hemithorax with tetracycline. Six months after the second pleurodesis, she was asymptomatic. ■

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