A rare case of lymphangioleiomyomatosis

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CASE

A 43-year-old Hispanic woman, a chronic smoker, presented with substernal chest pain associated with left arm numbness and headache. She also reported exertional dyspnea, dry cough, and night sweat for two months. Her oxygen saturation was 84% on room air. Lung examination revealed bilateral crackles. Her electrocardiogram showed a normal sinus rhythm with no signs of ischemia. Cardiac enzymes were negative. Computed tomography of the thorax with angiography showed no pulmonary emboli but numerous thin-walled cysts with variable sizes involving the entire lung bilaterally (Figures 1 and 2). No pulmonary nodules were identified. Her interferon gamma release assay was negative. Immunological workup, including P-ANCA, C-ANCA, anti-proteinase-3 (PR3) antibodies, anti-myeloperoxidase (MPO) antibodies, was negative. Vascular endothelial growth factor (VEGF-D) was normal at 36 pg/ml. Pulmonary function tests were normal with a FEV1 of 92% of predicted. Based on the unique CT images and clinical presentation, the patient was diagnosed with lymphangioleiomyomatosis (LAM). She was treated conservatively and did not require oxygen on discharge. The patient declined lung biopsy.

DISCUSSION

Lymphangioleiomyomatosis is a rare, idiopathic disorder that predominantly affects the lung parenchyma of women of childbearing age. It is characterized by bilateral pulmonary cystic changes on imaging and proliferation of abnormal smooth muscle cells on biopsy. The most common presenting symptoms are cough, dyspnea on exertion, and spontaneous pneumothorax.[1,2] Patients usually have a progressive loss of lung function. Radiological findings in the abdomen include chylous ascites, uterine leiomyomas, and abdominal and pelvic lymphangioleiomyomas. Renal angiomyolipomas develop in more than 50% of patients and may support the diagnosis suspected from the pulmonary presentation.[2] Affected patients may experience severe abdominal, flank, or pelvic pain, abdominal distention, incontinence, chyluria, hematuria, and lower-extremity lymphedema and paresthesias.

The radiologist is often the first clinician to suggest the diagnosis of LAM and might save the patient from unnecessary biopsy or surgical procedures. However, misdiagnosis is common and may result in inappropriate therapeutic procedures that can further complicate treatment.

Our patient had a normal serum VEGF-D level. However, this test has a low false- positive rate and a high false-negative rate, indicating that a positive result can be used to confirm LAM but a negative result should not be used to exclude LAM.[3] Disease severity and progression are variable and usually evaluated with pulmonary function and gas exchange testing, complemented by radiologic evaluation. Treatment measures include pleural interventions to control pneumothoraces, supplemental oxygen, kinase inhibitors, and lung transplantation.[3] Although survival rates for patients with LAM have improved over the past 2 decades, many experts continue to call for more objective data through therapeutic trials, which should be conducted prospectively with larger patient cohorts.

In summary, lymphangioleiomyomatosis is a serious progressive disease that predominantly affects women of childbearing age and leads to chronic incapacitating respiratory insufficiency. Clinical suspicion arises when diffuse cystic finding in CT images are found. Biopsy is rarely needed to confirm the diagnosis. At present, there is no curative treatment.

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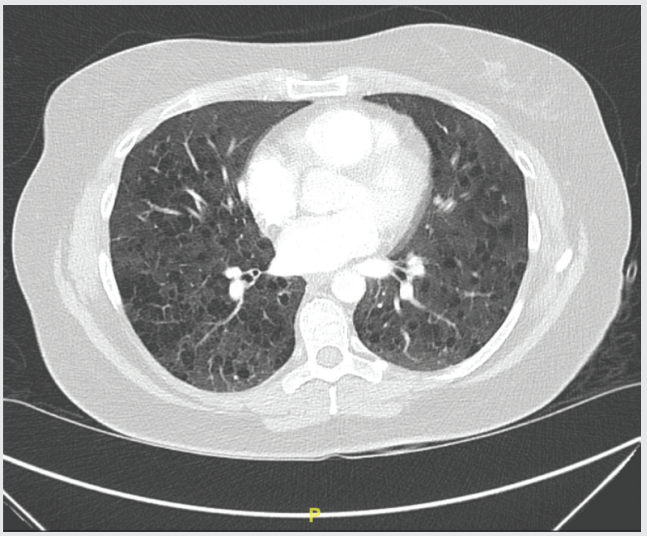

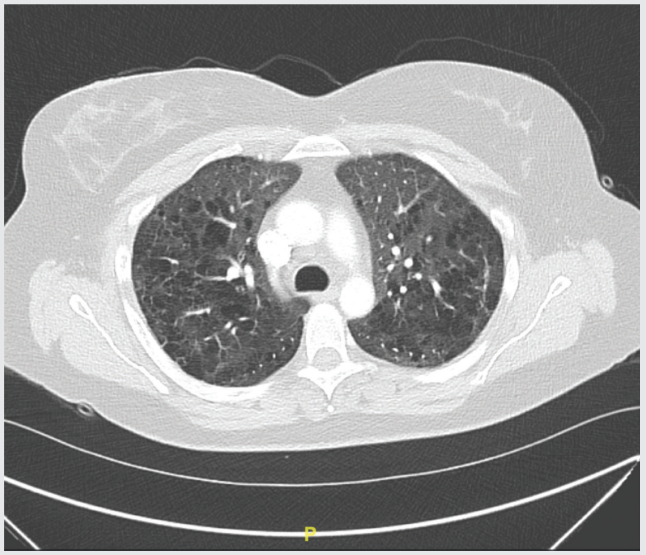

Figures 1 and **2.** Computed tomography scans demonstrate numerous thin-walled cysts with variable sizes involving the entire lung bilaterally.

Keywords: Lymphangioleiomyomatosis, cystic lung disease, diffuse lung disease

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References

1. Kalassian KG, Doyle R, Kao P, et al. Lymphangioleiomyomatosis: new insights. Am J Respir Crit Care Med 1997;155: 1183–1186.
2. Abbott GF, Rosado-de-Christenson ML, Frazier AA, et al. Lymphangioleiomyomatosis: radiologic-pathologic correlation. RadioGraphics 2005;25:803–828.
3. McCormack FX, Gupta N, Finlay GR, et al. ATS/JRS Committee on Lymphangioleiomyomatosis. Official American Thoracic Society/Japanese Respiratory Society clinical practice guidelines: lymphangioleiomyomatosis diagnosis and management. Am J Respir Crit Care Med 2016;194: 748–761.