Diagnostic Accuracy of Computed Tomography in Lymphangioleiomyomatosis

Valor Diagnóstico da Tomografia Computorizada na Linfangioleiomiomatose

Keywords: Lung Neoplasms/diagnostic imaging; Lymphangioleiomyomatosis/diagnostic imaging; Tomography, X-Ray Computed
Palavras-chave: Linfangioleiomiomatose/diagnóstico por imagem; Neoplasias do Pulmão/diagnóstico por imagem; Tomografia Computorizada

Dear Editor,

Lymphangioleiomyomatosis (LAM) is a rare disease classified by the World Health Organization (WHO) as a low-grade perivascular epithelioid tumor. It involves the development of multiple cysts in the epithelial layers due to the proliferation of abnormal smooth muscle cells, known as LAM cells.[1] This disease primarily affects women, typically around the age of 35, and while it is mostly idiopathic, it is associated with tuberous sclerosis in approximately 15% of cases.[2] Early symptoms are often mild, but the disease progresses and is characterized by dyspnea, with a strong association with spontaneous pneumothorax.[3] Additionally, about 1% of patients with scleroderma exhibit LAM-like changes in their lungs.[3]

We present the case of a 28-year-old woman with a 20-year history of systemic scleroderma who presented with shortness of breath lasting for two hours. Her medical history included reflux, gastritis, Raynaud's phenomenon, scoliosis, hands arthritis (interphalangeal), and nephrolithiasis. At the time, the patient was on daily omeprazole and amlodipine and received annual zoledronic acid treatment. She had no history of surgeries or other major diseases and had not experienced previous respiratory symptoms, despite being a long-term smoker. Muscle weakness was her initial symptom of systemic scleroderma. A thoracic computed tomography (CT) scan revealed multiple round, thin-walled cysts distorting her lung architecture (Fig. 1A). The CT also identified a pneumothorax, which was subsequently drained, as well as renal angiomyolipomas and nephrolithiasis (Fig. 1B). These CT findings led to a suspected diagnosis of lymphangioleiomyomatosis, which was confirmed through histopathological analysis of a transthoracic lung biopsy. The biopsy detected LAM cells, and immunohistochemical staining showed a positive reaction with the HMB-45 antibody. Unfortunately, the vascular endothelial growth factor-D (VEGF-D) test was not performed, as it was unavailable in the patient's city. The patient began treatment with sirolimus (2 mg/day), and due to significant lung function deterioration over the following month, she was placed on the lung transplant waiting list.

The diagnosis of LAM typically starts with non-invasive investigations, guided by clinical suspicion and characteristic findings on chest CT. To confirm the diagnosis, patients must also present at least one of the following: tuberous sclerosis complex, renal angiomyolipomas on CT or magnetic resonance imaging (MRI), lymphangioleiomyomas on abdominal or pelvic CT or MRI, chylous effusions, positive cytology for LAM cells in effusions or lymph nodes, or histopathological confirmation from a lung biopsy if needed. Elevated levels of VEGF-D, particularly in cases with lymphatic involvement, further support the diagnosis.[4]

Lymphangioleiomyomatosis is initially suspected based on chest CT scans, which typically reveal multiple cysts, usually 2 to 5 mm in diameter with thin walls. In severe cases, cysts may reach up to 12 mm and are usually distributed bilaterally.[2] A definitive diagnosis requires immunohistochemical staining and histopathological analysis,[3] which identify LAM cells by their spindle-shaped appearance, eosinophilic cytoplasm, and variable structures, ranging from nodules to small clusters. The nuclei are oval and contain fine chromatin.[2]

Treatment for LAM includes the use of bronchodilators to alleviate symptoms, which is effective in about a quarter of patients. Smoking cessation is also essential to delay disease progression.[5] Additionally, sirolimus has been shown to improve lung function and quality of life, as well as slow the progression of LAM.[2]

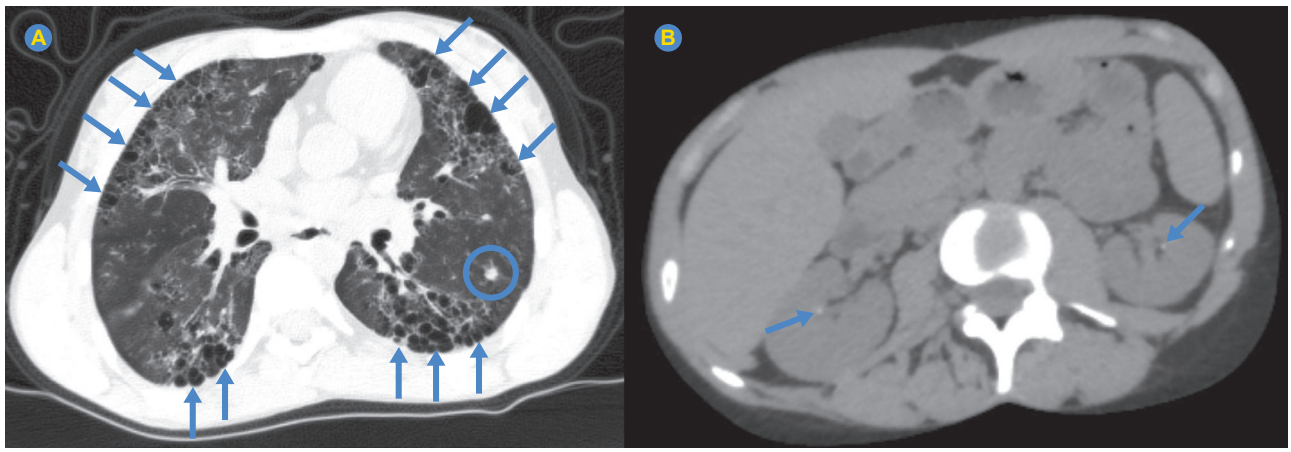

Figure 1 – Axial section of the patient's computed tomography (lung window), revealing multiple pulmonary cysts (arrows) and a nodule in the left lung (circle) (A). Abdominal computed tomography of the patient, demonstrating nephrolithiasis in both kidneys (arrows) (B).

AUTHOR CONTRIBUTIONS

All authors contributed equally to this manuscript.

PROTECTION OF HUMANS AND ANIMALS

The authors declare that the procedures were followed according to the regulations established by the Clinical Research and Ethics Committee and to the Helsinki Declaration of the World Medical Association updated in 2013.

DATA CONFIDENTIALITY

The authors declare having followed the protocols in use at their working center regarding patients' data publication.

PATIENT CONSENT

Obtained.

COMPETING INTERESTS

The authors have declared that no competing interests exist.

FUNDING SOURCES

This research received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

REFERENCES

1. Sekimoto Y, Suzuki K, Okura M, Hayashi T, Ebana H, Kumasaka T, et al. Uncommon radiologic computed tomography appearances of the chest in patients with lymphangioleiomyomatosis. Sci Rep. 2021;11:7170.
2. Sathirareuangchai S, Shimizu D, Vierkoetter KR. Pulmonary lymphangioleiomyomatosis: a case report and literature review. Hawaii J Health Soc Welf. 2020;79:224-9.
3. Cong CV, Anh TT, Ly TT, Duc NM. Pulmonary lymphangioleiomyomatosis (LAM): a literature overview and case report. Radiol Case Rep. 2022;17:1646-55.
4. Gupta N, Finlay GA, Kotloff RM, Strange C, Wilson KC, Young LR, et al. Lymphangioleiomyomatosis diagnosis and management: high-resolution chest computed tomography, transbronchial lung biopsy, and pleural disease management. an official American Thoracic Society/Japanese Respiratory Society clinical practice guideline. Am J Respir Crit Care Med. 2017;196:1337-48.
5. Johnson SR, Cordier JF, Lazor R, Cottin V, Costabel U, Harari S, et al. European Respiratory Society guidelines for the diagnosis and management of lymphangioleiomyomatosis. Eur Respir J. 2010;35:14-26.

Pedro Henrique SEGATT[1], Marcelo DE QUEIROZ PEREIRA DA SILVA[2], Thaís NOGUEIRA DANTAS GASTALDI[3], Márcio Luís DUARTE✉[1,3]

1. Universidade de Ribeirão Preto (UNAERP) - Campus Guarujá. Guarujá. Brazil.
2. WEBIMAGEM Telerradiologia. São Paulo. Brazil.
3. Diagnósticos da América S.A. (DASA). São Paulo. Brazil.

✉ **Autor correspondente:** Márcio Luís Duarte. marcioluisduarte@gmail.com

Recebido/*Received*: 25/07/2024 - **Aceite/*Accepted*:** 09/09/2024 - **Publicado *Online*/*Published Online*:** 23/09/2024 - **Publicado/*Published*:** 04/11/2024

https://doi.org/10.20344/amp.22116