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Case Report

Syndromatic Pulmonary Lymphangioleiomyomatosis

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Abstract

Lymphangioleiomyomatosis (LAM) is a rare disease that predominantly affects women, especially those of childbearing age. It occurs sporadically or is associated with tuberous sclerosis complex. The case of a 25-year-old female patient is presented, with a clinical history of fever and dry cough for one month evolution, as well as facial cutaneous angiofibromas, Shagreen patches on the neck and shoulders and Koenen nail tumors on the feet.

Keywords: Lymphangioleiomyomatosis; Tuberous Sclerosis; Cutaneous Angiofibromas; Shagreen Patches; Koenen's Tumors

Introduction

Pulmonary lymphangioleiomyomatosis represents a challenge in terms of its diagnosis, as well as in the management of its complications. Is a low-grade destructive metastasizing perivascular epithelioid cell tumors (PEComatous) resulting from the proliferation of lymphangioleiomyomatosis (LAM) cells in the lung, kidney and axial lymphatics. The disease is caused by mutations of the TSC2 or TSC1 genes and is more commonly sporadic rather than inherited [1]. Cystic lung disease (CLD) is the most frequent manifestation.

Clinical Case Presentation

A 25-year-old female patient, originally from San Antonio Huista, Huehuetenango, Guatemala, who consulted for fever and productive cough of 2 months' evolution. Within the medical history, refers to kidney failure diagnosed 5 years ago and without current treatment. On physical examination, multiple reddish lesions were observed on the nose and cheeks, in relation to facial cutaneous angiofibromas (Figure 1A). In the neck area and on the posterior part of the shoulder, multiple extensive plaques with an orange-peel appearance, yellowish-brown in color are observed, which are considered a type of connective tissue hamartoma in relation to Shagreen patches (Figure 1B and C). When exploring the left foot, reddish papules are observed that represent subungual and periungual fibromas, in relation to Koenen's tumors (Figure 1D).

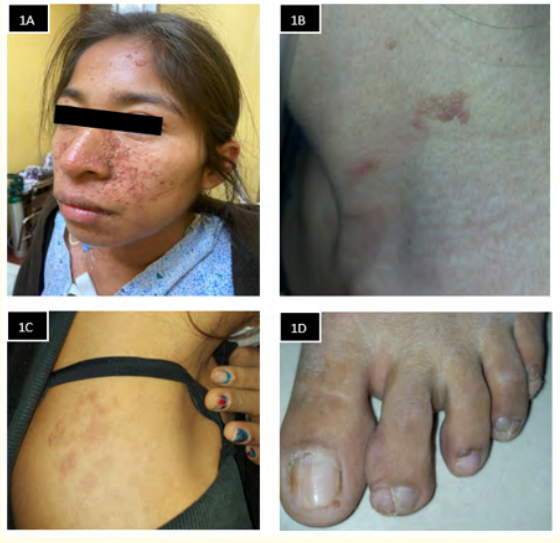


Figure 1: Clinical findings in Syndromatic pulmonary lymphangioleiomyomatosis. A) Facial cutaneous angiofibromas. B) Shagreen patches on the neck and C) on the posterior part of the shoulder. D) Nail Koenen tumors on the left foot.

The chest X-ray showed a diffusely distributed reticulonodular pattern in both lung fields (Figure 2). On physical examination he presented hepatomegaly, for which an abdominal ultrasound was requested, where multiple amorphous, hyperechogenic images were evidenced, diffusely distributed at the level of the right hepatic lobe, which are solid in appearance and with defined borders, the largest of them located at level of segment VII of the liver, which do not present vascular flow to the evaluation with color Doppler (Figure 3A), likewise, in projection of both renal fossae multiple amorphous, heterogeneous images are visualized, which deform the normal anatomy of both kidneys and cause an increase in the size of both kidneys, documenting what seems to correspond to the lower pole of the left kidney at the level of the pelvic cavity (Figure 3B). In the abdominal CT, the liver is observed enlarged, it has a heterogeneous appearance, secondary to the fact that at the level of the liver parenchyma and diffusely distributed, multiple images of different morphology are observed, hypodense on the periphery, with a hyperdense center, the which presents precontrast attenuation coefficients of -67 HU (Figure 4A). Both kidneys are visualized enlarged, occupying a large part of the abdominal cavity, the lower pole of both reaching the pelvic cavity, they present loss of their normal morphology, secondary to the fact that multiple hypodense amorphous images are visualized inside, with coefficients of -70 HU attenuation (Figure 4B). Said masses found in the liver and kidneys present a slight enhancement in the periphery, findings that together are related to angiomyolipomas (Figure 4C). Despite the bilateral enlargement of both kidneys, it is observed that both kidneys adequately concentrate the intravenous contrast medium, evidencing its excretion at the level of the urinary bladder (Figure 4D). Chest CT showed diffusely distributed multiple pulmonary parenchyma, rounded and homogeneous, with measurements ranging between 0.5 and 2.0 cm, thin-walled (<1 mm) and defined, highly defined findings, suggestive of pulmonary lymphangioleiomyomatosis (Figure 5 A and B). Subsequently, a brain tomography is performed, finding at the level of the subependymal space, multiple irregular images, hyperdense, with attenuation coefficients of up to + 898 HU, the largest of them located at the level of the body of the right lateral ventricle, measuring 0.8 x 0.6 cm, findings in relation to subependymal hamartomas (Figure 6 A and B).

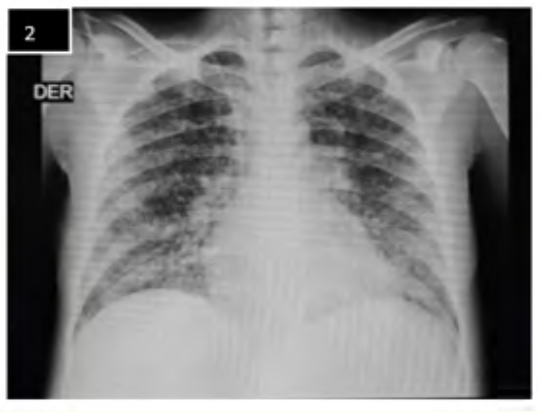


Figure 2: AP chest radiograph. Demonstrating a diffuse reticulonodular pattern involving both lung fields.

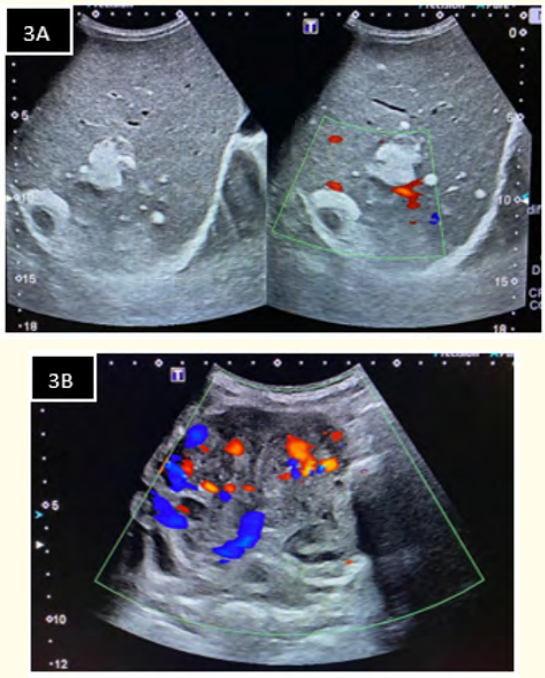


Figure 3: Ultrasonographic Findings of Syndromatic Pulmonary Lymphangioleiomyomatosis in the Abdomen. A) Multiple amorphous hyperechogenic images of diffuse distribution in the liver parenchyma, with a solid appearance and defined borders, without vascular flow in the color Doppler evaluation. B) multiple amorphous heterogeneous images that deform the normal anatomy of both kidneys and increase their size, visualizing the lower pole of the kidneys at the level of the pelvic cavity.

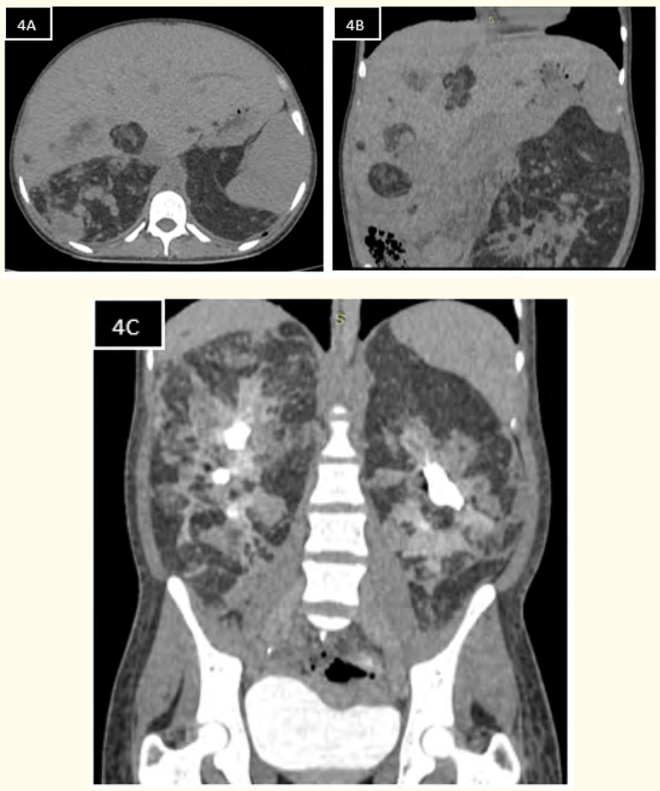
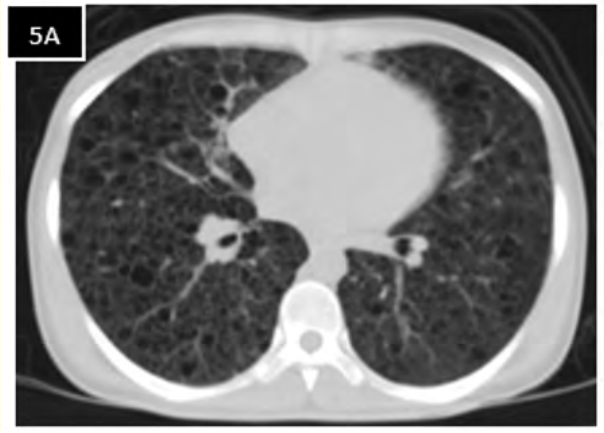


Figure 4: Abdominal CT findings associated with pulmonary lymphangioleiomyomatosis. A) Abdominal CT in axial section and single phase showing a lesion with a predominance of size at the level of the right hepatic lobe with attenuation coefficients of -70 HU and B) seen in coronal section. C) It can be seen that both kidneys concentrate and excrete the intravenous contrast medium, appreciating contrast in the urinary bladder.



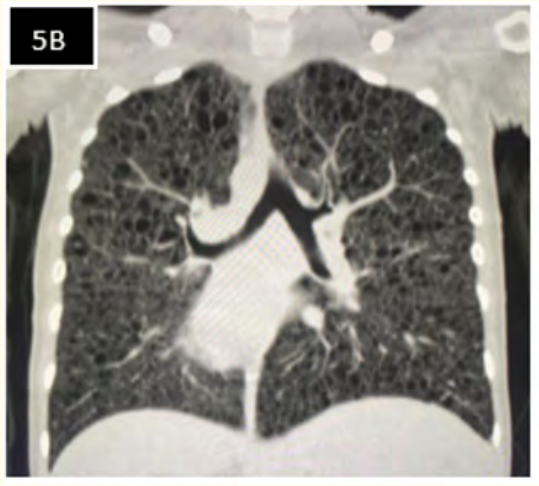


Figure 5: Pulmonary CT findings in Pulmonary Lymphangioleiomyomatosis. A) Pulmonary window and axial section, observing that throughout the lung parenchyma and diffusely distributed, multiple rounded and homogeneous pulmonary cysts, with thin and defined walls, measuring up to 2 cm, are observed. B) views in coronal sections.

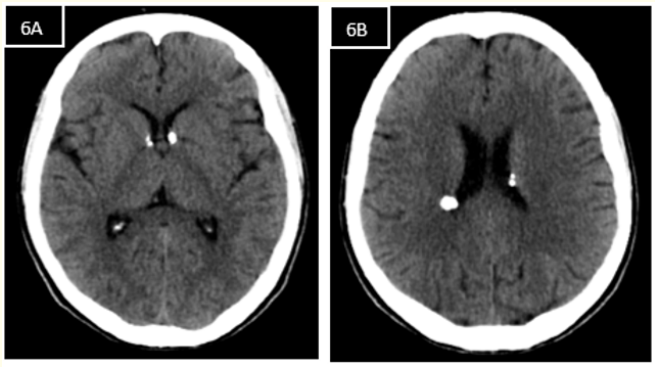


Figure 6: Findings of tuberous sclerosis in brain CT. A) and B) at the level of the subependymal space, multiple irregulars, hyperdense images are visualized, with attenuation coefficients of up to +898 HU, the largest of them at the level of the body of the right lateral ventricle, measuring 0.8 x 0.6 cm, findings in relation to subependymal hamartomas.

Discussion

Given the simplicity of the symptoms with which lymphangioleiomyomatosis can debut, its diagnostic confirmation occurs in advanced stages of the disease, when significant lung damage leads to the appearance of clinical factors with greater repercussions on the general condition of the patients, for which reason performing early imaging studies gains vital importance [2]. The present case is associated with findings of tuberous sclerosis. 50% of the patients have abdominal involvement; the most frequent are asymptomatic, multiple, bilateral renal angiomyolipomas [3].

Pneumothorax, progressive dyspnoea and chylous pleural effusions are the main clinical manifestations of lymphangioleiomyomatosis [4]. Dyspnoea is the most common symptom (over 70% of patients) and the result of airflow obstruction and cystic destruction of the lung parenchyma. Over 50% of patients have a history of pneumothorax in their clinical course. Other respiratory symptoms are cough, chyloptysis and haemoptysis. As described above, haemoptysis and chyloptysis may be the result of lymphangioleiomyomatosis cell obstruction of pulmonary blood vessels and lymphatics, respectively. Extrapulmonary manifestations of lymphangioleiomyomatosis are angiomyolipomas, which occur mostly in the kidneys, chylous ascites, abdominal lymphadenopathy and large cystic lymphatic masses termed lymphangioleiomyomas [5].

Recent guidelines recommend that patients with lymphangioleiomyomatosis or suspected lymphangioleiomyomatosis undergo baseline abdominal-pelvic CT to identify renal angiomyolipomas, enlarged lymph nodes, and lymphangioleiomyomas to plan management and follow the evolution of the disease [6]. The imaging findings of these abnormalities and the sequelae of their treatment may be confused with malignancies and other entities that may prompt unnecessary biopsies and surgical interventions [7]. Because the extent of lung disease may be severe and may result in decreased lung function, avoidance of surgery and other invasive procedures that might require ventilatory management is of particular importance in patients with lymphangioleiomyomatosis [8]. Currently the only curative treatment is lung transplant.

Conclusion

If the lung CT shows the characteristic cystic pattern, associated with an elevation of the vascular endothelial growth factor (VEGF), it would confirm the diagnosis of LAM. If there are doubts about the imaging pattern or the elevation of the VEGF is not obtained, it would be justified to obtain a lung biopsy. In order to obtain the diagnosis, given the prognosis of the disease [3]. For medical staff, LAM poses a challenge, both in diagnosis and management, due to its restrictive pattern, complications from episodes of pneumothorax, hemoptysis, and hemodynamic instability. LAM findings on CT are highly characteristic and can be considered diagnostic, particularly when typical abdominal lesions are also present.

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