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Retroperitoneal lymphangioleiomyoma with lymph node involvement: A pathologic-radiologic correlation of a rare form of myomelanocytic tumor

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ABSTRACT

Lymphangioleiomyomatosis (LAM) is a rare and slowly progressive disorder that usually arises in the lung, affects exclusively women in their childbearing years, and typically presents with progressive dyspnea on exertion and pneumothorax. Infrequently, extra-pulmonary LAM can occur in the retroperitoneum, uterine wall, mediastinum and intraperitoneal lymph nodes. Histologically, LAM is characterized by a proliferation of perivascular epithelioid cells (PEC) that express markers for both melanocytes and smooth muscle cells.

We report a case of a peripancreatic retroperitoneal mass that was incidentally discovered on magnetic resonance image (MRI) scan of a 38-year-old female. The morphologic findings and the immunohistochemical staining were consistent with a lymphangioleiomyoma. The radiologic and pathologic correlation along with differential diagnosis of this rare entity is discussed.

1. Introduction

Lymphangioleiomyomatosis (LAM) is a rare disorder with an estimated incidence of 1 in 1 million people in the United States and Europe [1]. It is a pulmonary disorder that exclusively affects young women of childbearing age. The development of LAM both in inherited and sporadic forms is related to loss of function mutations of gene TSC2 which is one of the two loci involved in the pathogenesis of tuberous sclerosis. TSC2 encodes for a tuberin protein, a negative regulator of the mammalian target of rapamycin (mTOR). Stimulation of mTOR is associated with cell growth and proliferation. The loss of heterozygosity (LOH) of TSC2 and the associated increased mTOR activity makes the latter a reliable target for therapeutic applications [2].

LAM has rarely been described to involve the retroperitoneum and mediastinum [3]. The disease is characterized by a proliferation of morphologically distinctive spindle cells that express reactivity for both smooth muscle and melanocytic markers [4]. Extrapulmonary presentation of LAM as a discrete, solitary tumor mass (lymphangioleiomyoma) is exceedingly rare and may present challenges for histologic and radiographic diagnosis.

2. Case report

A 38-year-old Caucasian woman volunteered to be part of a magnetic resonance image (MRI) screening exam at her work. She was

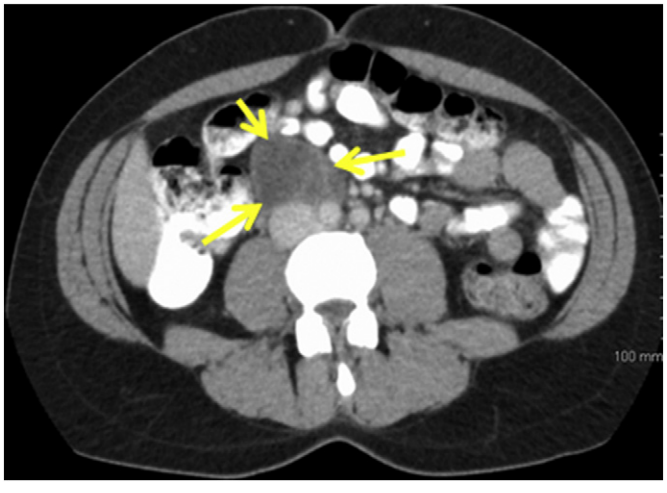

Fig. 1. Axial contrast enhanced computed tomography scan demonstrating a hypoattenuating, septated retroperitoneal mass (yellow arrow) anterior to the aorta and inferior vena cava.

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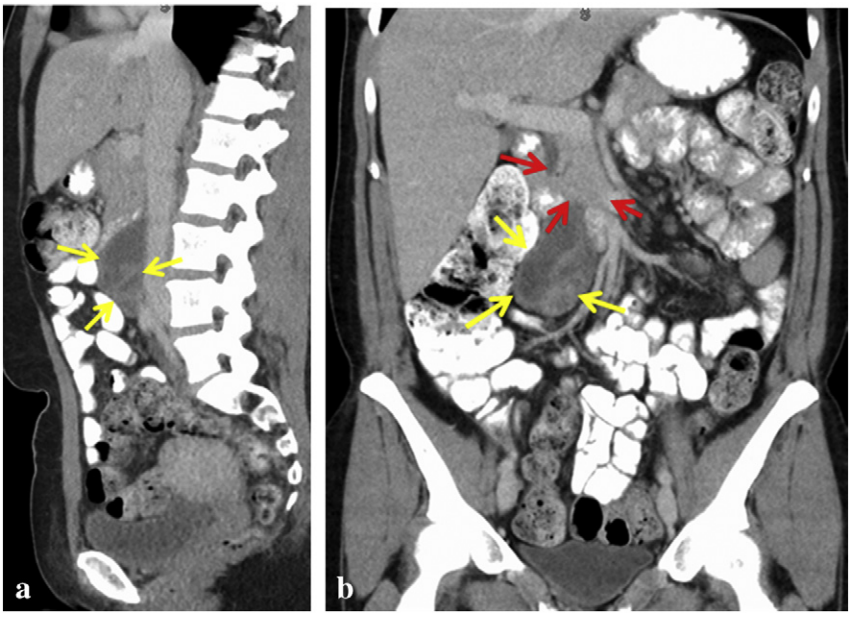

Fig. 2. (a) Coronal (left) and (b) sagittal (right) contrast enhanced CT scans demonstrate a hypoattenuating partially cystic retroperitoneal mass (yellow arrows) which abuts the pancreatic head (indicated by red arrows on the right figure).

incidentally found to have a peripancreatic mass in the right retroperitoneal region. She had a history of chronic right lower quadrant pain that radiated to her right flank and back for approximately 7 years that had been thought to be of gynecologic origin. The patient had a history of smoking and complained of cough for over 10 years without dyspnea. The patient also had hypothyroidism, atopic dermatitis, and chronic sinusitis. Past medical history included tonsillectomy, adenoidectomy, anterior cruciate ligament reconstruction, and two cesarean sections. Her family history is significant for breast cancer in her mother and grandmother, prostate cancer in her father, and lung cancer and lymphoma in her maternal grandfather. There is no family history of tuberous sclerosis (TSC).

3. Radiologic features

Abdominal computed tomography (CT) (Figs.s 1, 2) scan showed a partially cystic and partially solid mass that appeared to abut the inferior margin of the head of the pancreas; however it did not appear to arise from the pancreas. The mass measured approximately 6.9 cm in craniocaudal dimension, 4.6 cm in transverse dimension and 3.8 cm in anterior-posterior dimension. Margins of the mass were relatively smooth and circumscribed.

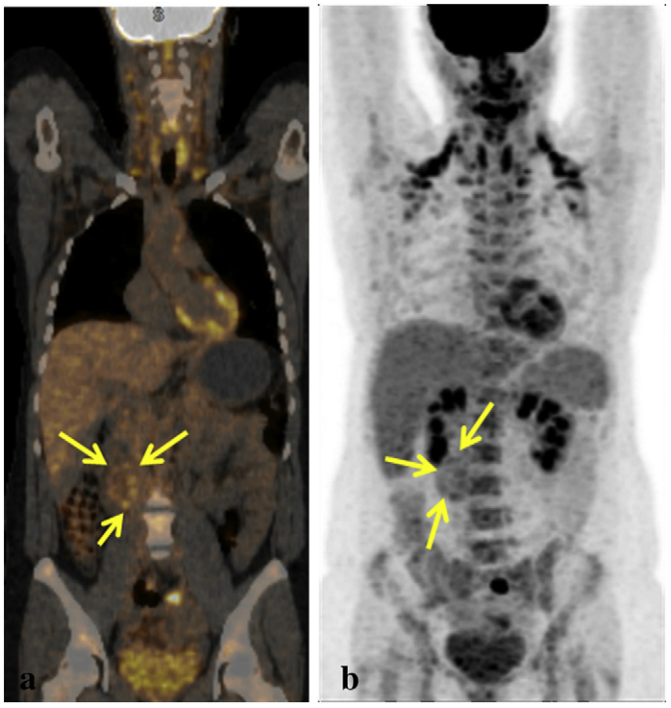

Fig. 3. (a, b). Coronal PET images demonstrating mild FDG uptake in the retroperitoneal mass (yellow arrows).

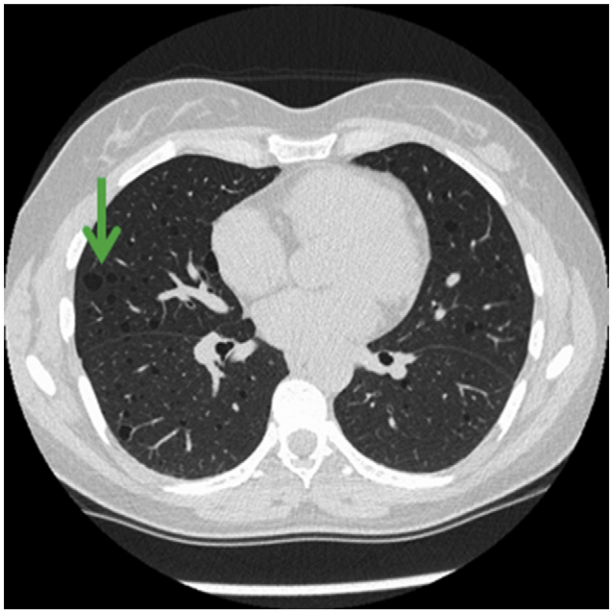

Fig. 4. Axial chest CT image reveals multiple well circumscribed cysts throughout both lungs (green arrow).