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ΑΡΧΕΙΑ ΕΛΛΗΝΙΚΗΣ ΙΑΤΡΙΚΗΣ 2017, 34(1):134–135

Medical Imaging Quiz – Case 42

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A 30-year-old female presented with aggravated dyspnea and cough. She had given birth to her first baby three months ago. Physical examination showed reduced pulmonary sounds on the right side. Chest X-ray confirmed the diagnosis of right-sided pneumothorax. Computed tomography (CT) revealed reticular interstitial pattern and multiple bilateral cysts, pneumothorax of the right lung and small pleural effusion. The diagnosis was confirmed by pulmonary biopsy.

Comment

Lymphangioleiomyomatosis (LAM) is a rare, cystic lung disease that primarily affects young women. Exacerbations of LAM have been reported to occur during pregnancy and menstruation, as well as with exogenous estrogen use. The most common presentation of LAM is progressive dyspnea on exertion (difficulty breathing with physical activity), often in association with a history of pneumothorax or chylothorax. LAM occurs in two settings: In the disease tuberous sclerosis complex (TSC-LAM), an autosomal dominant disorder due to mutations in the TSC1 or TSC2 genes and in a sporadic form, in women who do not have TSC (sporadic LAM or S-LAM). Because many of the early signs and symptoms of LAM are similar to those of other lung diseases, including asthma, emphysema and bronchitis, LAM can be difficult to diagnose. On average, women with LAM have symptoms for 3 to 5 years and suffer an average of 2.2 pneumothoraces before the diagnosis of LAM is made.

There are a number of tests that can confirm or rule out the existence of LAM and evaluate the extent of the disease or of lung damage.

These include: Patient profile involves obtaining a full history, including smoking, use of birth control pills, seizure history and family history of TSC. Other manifestations of LAM include renal angiomyolipomas, chylous effusions, lymphangioleiomyomas.

Chest radiographs in LAM may be normal. Fine reticular or reticulonodular interstitial infiltrate with preserved lung volumes is the most commonly observed abnormality. Pleural effusions may be present. Patients may present with pneumothorax.

High-resolution CT (HRCT) scan is the most accurate imaging test for diagnosing LAM, providing a correct diagnosis more than 80% of the time. The features of chest CT are nearly pathognomonic with the detection of diffuse thin-walled cysts, adenopathy and thoracic duct dilatation, pleural effusion, pneumothorax, ground-glass opacities, pericardial effusion and multifocal multinodular pneumocyte hyperplasia.

HRCT is recommended for patients with known TSC: All females, at least once after the age of 18 and all patients who present with pneumothorax or chylothorax. For patients who do not have known TSC: all non-smoking women who present with spontaneous pneumothorax, all women who present with recurrent spontaneous pneumothorax regardless of smoking status, and all women found to have an angiomyolipoma, abdominal lymphangiomyoma or chylous effusion.

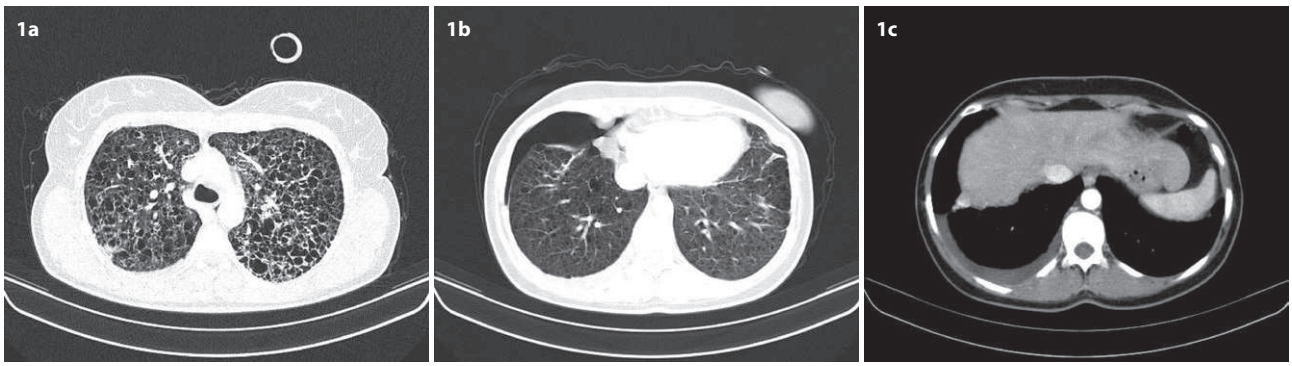

Figure 1. Computed tomography revealing reticular interstitial pattern and multiple bilateral cysts (a), pneumothorax of the right lung (b), and small pleural effusion (c).

In the presence of compatible CT findings, and the presence of any one of the following (TSC, angiomyolipomas, chylous effusions, lymphangioleiomyomas, histologically confirmed LAM from enlarged lymph nodes, elevated serum vascular endothelial growth factor-D [VEGF-D] greater than 800 pg/mL) lung biopsy can be skipped. In the absence of the above mentioned confirmatory features, the diagnosis of LAM requires a lung biopsy for confirmation. Lung tissue can be acquired by thoracoscopy (video-assisted thorascopic surgery, VATS) or transbronchial biopsy. The amount of tissue obtained by transbronchial biopsy is less than in a VATS procedure and may not be adequate to definitively diagnose LAM.

VEGF-D blood-based diagnostic test can distinguish LAM from other cystic lung diseases that present with similar HRCT scan appearances. In patients with a compatible HRCT chest, a high VEGF-D value (greater than 800 pg/mL) is diagnostic for LAM, and no other confirmatory test is needed in that scenario. Thus, a high VEGF-D level can save the need for an invasive lung biopsy to diagnose LAM. It is important to remember, however, that a normal VEGF-D level does not rule out the diagnosis of LAM. VEGF-D may also be useful as a screening test for LAM in women with TSC.

Treatment strategies presently available for LAM are based on inhibition of the mechanistic target of rapamycin (mTOR) pathway. The use of sirolimus is now FDA approved for treatment of patients with LAM who have abnormal lung function and evidence of progressive lung function decline, or with refractory chylous effusions. A trial of bronchodilators is often used with LAM patients who have a reversible airflow obstruction based on pulmonary function testing.

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