

Spontaneous Pneumothorax in a Young Female with Lymphangiomyomatosis

Lymphangiomyomatosis (LAM) is a rare disorder of abnormal proliferation of smooth muscle cells which results in the formation of thin-walled cysts and progressive lung destruction. It is a multi-organ disease that insidiously affects females of reproductive age. We present a new onset LAM in a young female with hydropneumothorax.

Case: A 33-year-old woman with no past medical history presented to the emergency department with sudden onset pleuritic chest pain while pushing her child's stroller. The pain improved with rest but recurred severely while at work prompting her to visit the hospital. She denied similar episodes, recent trauma, or lung disease. In the emergency she appeared uncomfortable, with decreased left-sided breath sounds. Chest pain was worse with left arm movement. CBC, CMP, APTT & D-Dimer were within normal limits & COVID was negative. Chest X-ray revealed a large left-sided hydropneumothorax with a mediastinal shift to the right. Pigtail catheter was placed. CT imaging to investigate secondary causes of pneumothorax showed thin-walled cysts diffusely throughout the lungs with ground glass background suspicious for LAM. Left video-assisted thoracoscopy (VATS) with left lung biopsy was performed by cardiothoracic surgery with biopsy results consistent with proliferation of abnormal smooth muscle cell and HMB-45 staining confirmed the diagnosis of LAM. She was discharged with pulmonary follow up and initiated on Sirolimus.

LAM can be suspected in women of reproductive age, presenting with pneumothorax on chest X-ray without otherwise plausible etiology. LAM typically presents as diffuse thin-walled cysts surrounded by normal lung parenchyma on CT imaging. Lung biopsy may demonstrate the proliferation of atypical smooth muscle-like cells, normal lung architecture distorted by multiple small cysts and HMB-45 staining positivity. Physicians should be aware of such a rare condition, particularly in an at-risk population presenting with pneumothorax.