

Erdheim-Chester Disease as a Rare Cause of Recurrent Pleuro-Pericarditis

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Abstract

Erdheim-Chester disease is a rare non-Langerhans cell histiocytic neoplasm that can involve multiple body organs, mostly the bones. We hereby present a unique case of recurrent pericarditis caused by Erdheim-Chester disease diagnosed three years after the first episode of pericarditis.

A 54-year-old female from Peru with history relevant for acute pericarditis, peripheral neuropathy and interstitial lung disease presented to the emergency department for acute onset pleuritic chest pain, shortness of breath and orthopnea. Three years ago, she had an episode of acute pleuropericarditis, for which she underwent pericardiocentesis and thoracentesis which were nondiagnostic. She was diagnosed with acute idiopathic pericarditis. Since then, she has been having progressive shortness of breath with exertion. The patient was hemodynamically stable. On physical examination, she had jugular vein distention, Kussmaul sign, pericardial rub, hepatomegaly and peripheral edema. Laboratory studies were relevant for white blood cell count of 16,460 k/uL (normal: 3.70 - 11 k/uL), erythrocyte sedimentation rate of 48 mm/hour (normal: 10- 20 mm/hour) and C-reactive protein of 17.3 mg/dL (normal: <0.9 mg/dL). An echocardiogram done showed moderate circumferential pericardial effusion with constrictive physiology concerning for effusive-constrictive pericarditis. A pericardial window was then attempted, but it was unsuccessful because of the thick pericardium and pericardial adhesions. She was discharged on furosemide 20 mg orally daily and prednisone 60 mg orally twice a day. Her symptoms partially improved; however, she continued to have dyspnea with moderate exertion. A follow up cardiac magnetic resonance imaging a month later showed trivial pericardial effusion with severe diffuse circumferential pericardial thickening of 12 mm, moderate circumferential pericardial enhancement and diffuse pleural enhancement bilaterally (see attached image). The findings were consistent with persistent pericarditis, so she underwent a radical pericardiectomy and lung biopsy. Surgical pathology of the pericardium and the lung revealed diffuse infiltration of histiocytes consistent with Erdheim-Chester disease and associated with BRAF-V600E mutation. Following hospital discharge, the patient was prescribed vemurafenib which is a BRAF Inhibitor recently FDA approved for this condition. The patient's symptoms and exercise tolerance gradually improved to this date.

This case illustrates an example of a rare systemic disease causing recurrent pericarditis, recurrent pleural effusion and interstitial lung disease. With less than 500 reported cases of Erdheim-Chester worldwide, and few of them having cardiac involvement, the diagnosis can be challenging and most often delayed. Unfortunately, cardiac involvement in this disease has been reported to carry a poor prognosis, especially without targeted treatment. Plus, the etiology of recurrent pericarditis is advised to be addressed early on to decrease the risk of constrictive pericarditis and prolonged steroid use. About half of patients with Erdheim-Chester disease have positive BRAF-V600E mutation for which vemurafenib is efficacious in many cases based on the scarce data in the literature. In conclusion, in the presence of an unexplained multiorgan involvement (like pericardial effusion, pleural effusion, and interstitial lung disease), a pericardial and lung biopsy should be considered to diagnose rare systemic diseases, like Erdheim-Chester, and initiate early targeted treatment to decrease morbidity and

mortality.



Cardiac magnetic resonance imaging showing circumferential involvement (arrows).

Categories

Resident Case

Program Name

Cleveland Clinic, Fairview Hospital Internal Medicine residency program