Erdheim-Chester Disease

Alexander Poellinger, MD, and Joris Hrycyk, MD

From Department of Diagnostic, Interventional and Pediatric Radiology, Inselspital Universitatsspital Bern, Switzerland.

A 58-year-old man presented to the emergency department with cardiac tamponade and dilated cardiomyopathy of unknown origin. Chest radiography demonstrated cardiomegaly and pleural effusion. Computed tomography scan revealed pericardial masses of soft tissue-density between the atria and between the right atrium and ventricle (Figure 1); pericardial effusion; pleural effusion; enlarged and ill-defined adrenal glands; perirenal stranding (Figure 2); and sclerosed osseous lesions in the diaphysis of both humeri, both femoral necks, and the left os ilium. A biopsy taken from the femoral neck confirmed the suspected diagnosis of Erdheim-Chester disease. Erdheim-Chester disease is a rare form of non-Langerhans cell histiocytosis, which frequently involves multiple organ systems and responds poorly to therapy.

Erdheim-Chester disease is an important differential diagnosis for every clinician because of its varying appearance in multiple organ systems. Because secondary cardiac tumors are much more common than primary cardiac tumors like myxoma, cardiac tumors should make the clinician look for other tumors and disease manifestations, respectively.

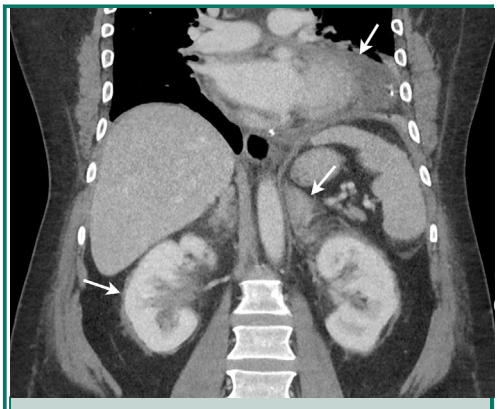

FIGURE 2. Pericardial effusion (**arrow**), pleural effusion, enlarged and ill-defined adrenal glands (**arrow**), and perirenal stranding (**arrow**).

The combination of cardiac tumors with various lesions in multiple organ systems should make one consider Erdheim-Chester disease as a differential diagnosis. Although it is a rare disease, increasing numbers in the last few years suggest that it is probably underdiagnosed because of previous unawareness of the disease.

Approximately 550 cases have been reported worldwide. Fifty percent of patients

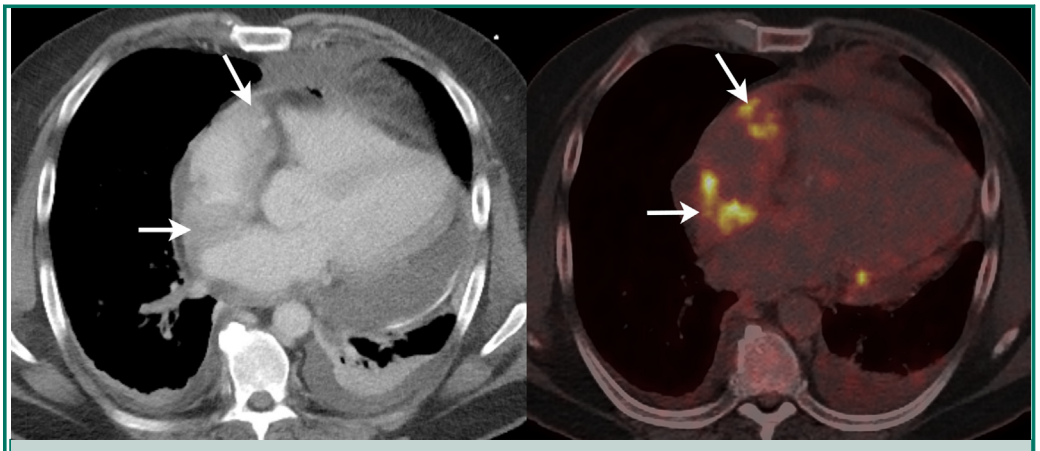

FIGURE 1. Pericardial masses of soft tissue-density between the atria and between the right atrium and ventricle (**arrows**).

Mayo Clin Proc. ■ May 2019;94(5):924-925 ■ https://doi.org/10.1016/j.mayocp.2019.01.026
www.mayoclinicproceedings.org ■

test positive for the BRAF V600E mutation. One month after the diagnosis, the patient presented with increasing shortness of breath and weight gain. He is currently undergoing treatment with interferon-alpha.

Potential Competing Interests: The authors report no competing interests.

Correspondence: Address to Joris Hrycyk, Department of Diagnostic, Interventional and Pediatric Radiology, Inselspital Universitatsspital Bern, Freiburgstrasse 10, Bern 3010, Switzerland (joris.hrycyk@insel.ch).