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Letter to the Editor

The Spectrum of Cardiac Involvement in Erdheim-Chester Disease is Broader than Previously Thought

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We read with interest the article by Wong *et al*. about two patients with Erdheim-Chester disease (ECD) with cardiovascular involvement presenting as encased aorta and its branches, coronary arteries, pulmonary arteries, and both atria (patient-1, 24yo female) as well as left renal artery stenosis, inferior and superior mesenteric artery stenosis, and stenosis of the celiac trunk (patient-2, 68yo male) [1]. Patient-1 was diagnosed with ECD based on histiocytes positive for CD68, CD163, and S100, and presence of a *MAP2K1* mutation [1]. Patient-2 was diagnosed with ECD due to stenosis of the superior and inferior mesenteric arteries, coeliac trunk, left renal artery, occlusion of the right renal artery, hairy kidneys, and retroperitoneal fibrosis [1]. The study is impressive, but some points require further discussion.

The first point is that the list of cardiac manifestations of ECD is incomplete and not only includes pericardial thickening, myocardial thickening, atrial pseudotumor, infiltration of the atrio-ventricular groove, atrial pseudotumor, coronary artery stenosis or occlusion which manifest with angina chest pain or myocardial infarction, vena cava infiltration, and supraventricular or ventricular arrhythmias [1]. One of the cardiac manifestations not taken into account is myocarditis. Although rarely reported, it needs to be included in the list of cardiac manifestations of ECD because it can greatly influence the outcome of ECD patients. This is because myocarditis can be complicated by heart failure and malignant ventricular arrhythmias. In a 67-year-old male with ECD with cardiovascular involvement (pathological tissue surrounding the atrioventricular sulcus and both cardiac atria, extending to the superior vena cava, pulmonary trunk, and ascending aorta, apical akinesia, reduced ejection fraction, and myocardial oedema) myocarditis was reported for the first time [2]. Endo-myocardial biopsy in this patient revealed interstitial edema, myocyte necrosis, focal lymphomonocyte infiltrate, and CD68+ foamy macrophages [2]. We also reported a 68-year-old female with ECD in whom late gadolinium enhancement (LGE) and pericardial effusion suggested myocarditis and pericarditis [3].

Endocarditis as a manifestation of ECD is also not mentioned. In a patient with ECD who underwent Ross procedure for suspected endocarditis, examination of the explanted pulmonary homograft confirmed endocarditis [4]. There is also a report of an ECD patient with pulmonary hypertension, who additionally manifested with arterial hypertension, heart failure, and hairy kidneys [5]. This particular patient benefited from methylprednisolone and alpha-interferon [5].

A second point is that the diagnosis of ECD in patient-2 is not certain. The patient had stenosis of the right renal artery, the superior and inferior mesenteric artery, and celiac trunk, occlusion of the left renal artery, and soft tissue around both kidneys, renal sinuses, and the uretero-pelvic junction [1]. However, histological examination did not show foamy CD68, C163m or S100 positive histiocytes and the patient was not examined for *BRAF*, *KRAS*, *NRAS*, *PIK3CA*, or *MAP2K1* mutations [6].

The third point is that the *BRAF* V600E mutation is not determined by immunehistochemical staining, as mentioned in the description of patient-2, but by molecular genetic testing. This should be corrected.

A fourth point is that "pericardial window" is the same as a "pleuro-pericardial" window [1]. There is no need to distinguish the two.

In summary, the interesting study has limitations that put the results and their interpretation into perspective. Clarifying these weaknesses would strengthen the conclusions and could improve the study. The spectrum of cardiac involvement in ECD is broader than expected and the diagnosis of ECD should be based not only on the clinical presentation but also on histological and immunehistological examinations as well as genetic testing.

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