

**Control Number:** 38

**Abstract Category:** Clinical Case Challenge in Cardio-Oncology

**Title:** Cardiac Tamponade as the first manifestation of Erdheim-Chester Disease: a rare case in literature

## **ABSTRACT BODY**

### **Background and Purpose**

Erdheim-Chester Disease (ECD) is classified as inflammatory myeloid neoplasia with an unknown incidence (1, 2). In general, it is multisystemic, include the long bones, skin, tissues behind the eyeballs, lungs, brain, pituitary gland, and/or additional tissues and organs (1, 2). The most frequent symptom is bone pain (2). Cardiovascular involvement is common but frequently asymptomatic and it is present in more than half of the patients; with infiltration of the aorta and atrial pseudotumor being the most common forms (1, 2). We present a rare case of symptomatic pericardial disease as first clinic presentation of ECD.

### **Case Description and Outcomes**

A 55-year-old female patient presented progressive dyspnea to minimum efforts, orthopnea and limb edema within 3 months. She evolved with short-term syncope and sphincter release. She was taken to an emergency department. There, she underwent to a transthoracic echocardiography (TTE), which found signs of cardiac tamponade. Pericardial drainage with pericardial fluid analysis, bacterial cultures, fungi culture, and AARB tests were performed; all of them were negative. Pericardial biopsy was also performed, which showed no changes. Therefore, an etiological investigation was carried out with cardiac magnetic resonance imaging (CMRi), which detected: right atrium (RA) with enlarged dimensions; presence of irregular contoured tissue formation infiltrated in RA wall, affecting the interatrial septum, aorta, right coronary artery and vena cava; discrete perfusion and heterogeneous late gadolinium enhancement; presence of suggestive image of thrombus in the RA; absence of myocardial fibrosis; such findings on CMRi are suggestive of non-Langerhans cell histiocytosis, which are characteristic of ECD (Figure 2). Finally, in order to confirm the diagnosis, the adrenal biopsy evidenced histiocytic proliferation, confirming the diagnosis of ECD, and PET-CT 18 FDG had hypercaptation in the middle mediastinal region and bones, confirming the diagnosis of ECD (Figure 3).

### **Discussion**

As the best of our knowledge, this is the first case reported in the literature which evidence a cardiac tamponade as initial manifestation of ECD. The ECD is a rare histiocytic disorder. The histiocytic disorders have been divided into Langerhans cell and non-Langerhans cell histiocytosis. The first one is so named for its presumed derivation from the Langerhans cells, which are specialized dendritic cells found in the skin and mucosa. In contrast, non-Langerhans histiocytosis are thought to be derived from the monocyte-macrophage lineage (3). The mean age at diagnosis is 53 years and there is a slight male predominance (4). The pathogeny is not clear, but a BRAF V600E mutation is related (2, 4). The diagnosis of ECD is made by identifying distinctive histopathological findings in the appropriate clinical and radiologic context (2). A circumferential soft-tissue sheathing of the thoracic and abdominal aorta is present about 56% and a mural pseudo-tumoral infiltration of the RA is present in up to 30% of patients, visualized clearly on MRI (1, 2). Pericardial disease occurs in 40% to 45% of patients, manifesting as pericarditis, effusion, and even cardiac tamponade (1, 2). Renovascular hypertension may develop when renal arteries are involved and may require treatment with stent. Coronary arterial disease can result in myocardial infarction. Diffuse infiltration of the myocardium or interatrial septum has been described, occasionally leading to heart failure (2,5, 6). The cardiac involvement in ECD gives a worse prognosis (7). Treatment involves

interferon- $\alpha$ , corticosteroids, cytotoxic chemotherapies, serine/threonine kinase inhibitors (vemurafenib and imatinib), radiotherapy, and surgery (2).

## References

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Image 1

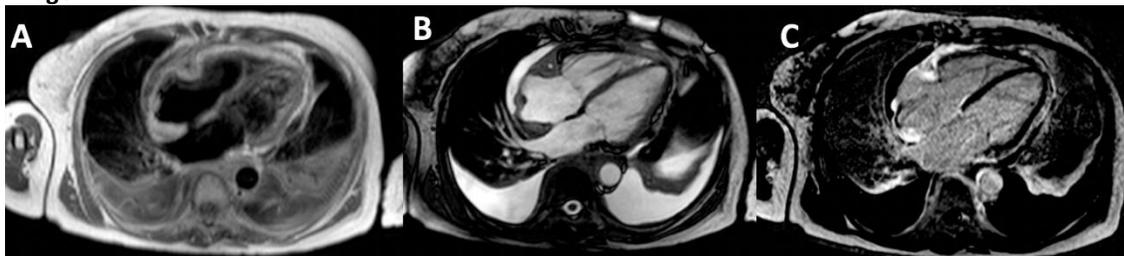


Image 2

