

## BACKGROUND

- Erdheim-Chester disease (ECD) is classified as an inflammatory myeloid neoplasia. This is a multisystemic, affecting long bones, skin, tissues behind the eyeballs, lungs, the brain, the pituitary gland, and other tissues and organs.
- Cardiovascular involvement is common, present in more than half of patients but frequently asymptomatic. However, it is associated with a worse prognosis.

## CASE REPORT

- A 55-year-old woman, presented progressive dyspnea to minimum efforts, orthopnea, atypical chest pain and limb edema in the last 3 months.
- In her initial evaluation the blood pressure was 80/60 mmHg and electrocardiography showed sinus tachycardia with low QRS voltage.
- The transthoracic echocardiography (TTE) in an emergency department detected signs of cardiac tamponade – Figure 1.
- Surgical pericardiocentesis was performed with drainage of 600 ml of serous fluid. The fluid was compatible with inflammatory transudate and no malignant cells were found. The biopsy showed slight inflammatory pericarditis.
- The cardiac magnetic resonance (CMR) detected characteristic findings of ECD like presence of irregular contoured tissue formation infiltrated in RA wall, affecting the interatrial septum, aorta, right coronary artery and vena cava, discreet perfusion and heterogeneous late gadolinium enhancement; presence of suggestive image of thrombus in the RA - Figure 2.
- The adrenal biopsy demonstrated histiocytic proliferation, with positivity for CD68, CD163, S-100 protein (focal), BRAF and negativity for CD1A.
- The 18-Fluorodeoxyglucose positron emission tomography-computed tomography (18-FDG PET-CT) showed bone and thoracic involvements characterized by high uptake (SUV max: 6.3 on femur) in the middle mediastinal region and bones – Figure 3.
- The treatment with interferon and zoledronic acid was started.

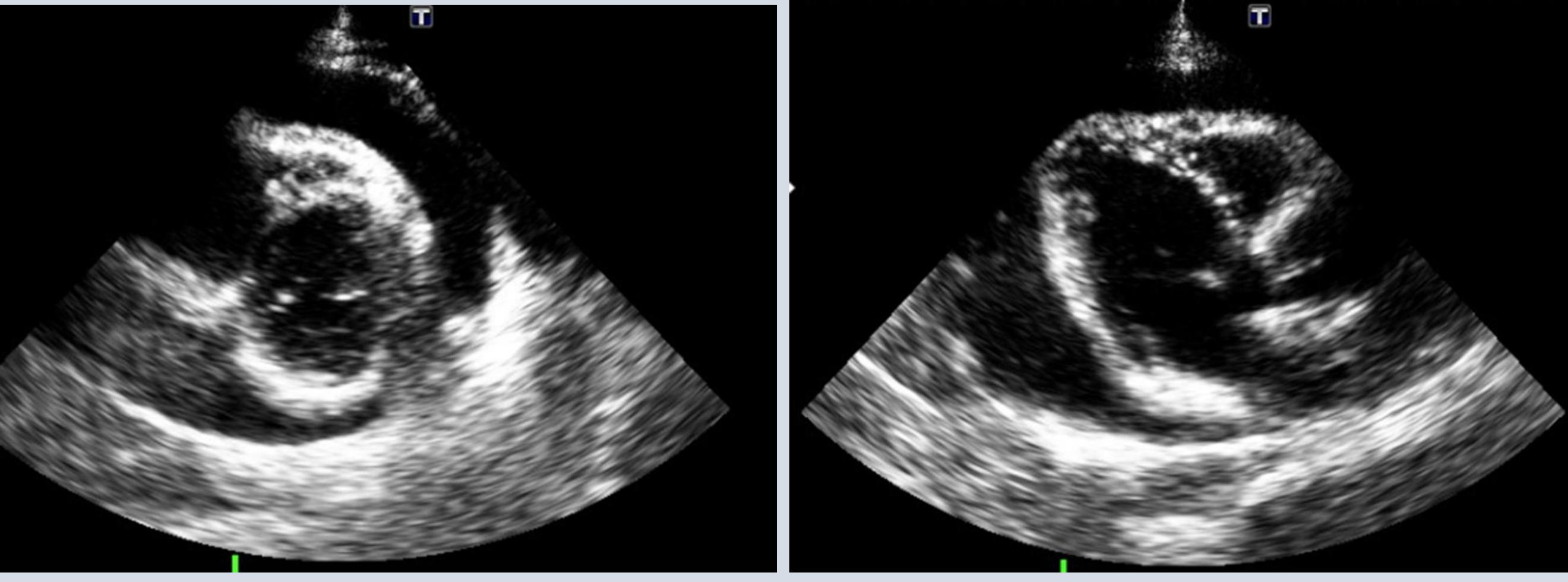


Figure 1 – ECHO: Showed signs of cardiac tamponade.



Figure 2 – CMR: (A) Cine 4 chambers - presence of irregular contoured tissue formation infiltrated in RA wall, affecting the interatrial septum, aorta, right coronary artery. (B) Cine 4 chambers - discreet perfusion and heterogeneous late gadolinium enhancement are observed. (C) 4 chambers – presence of edema transmural.

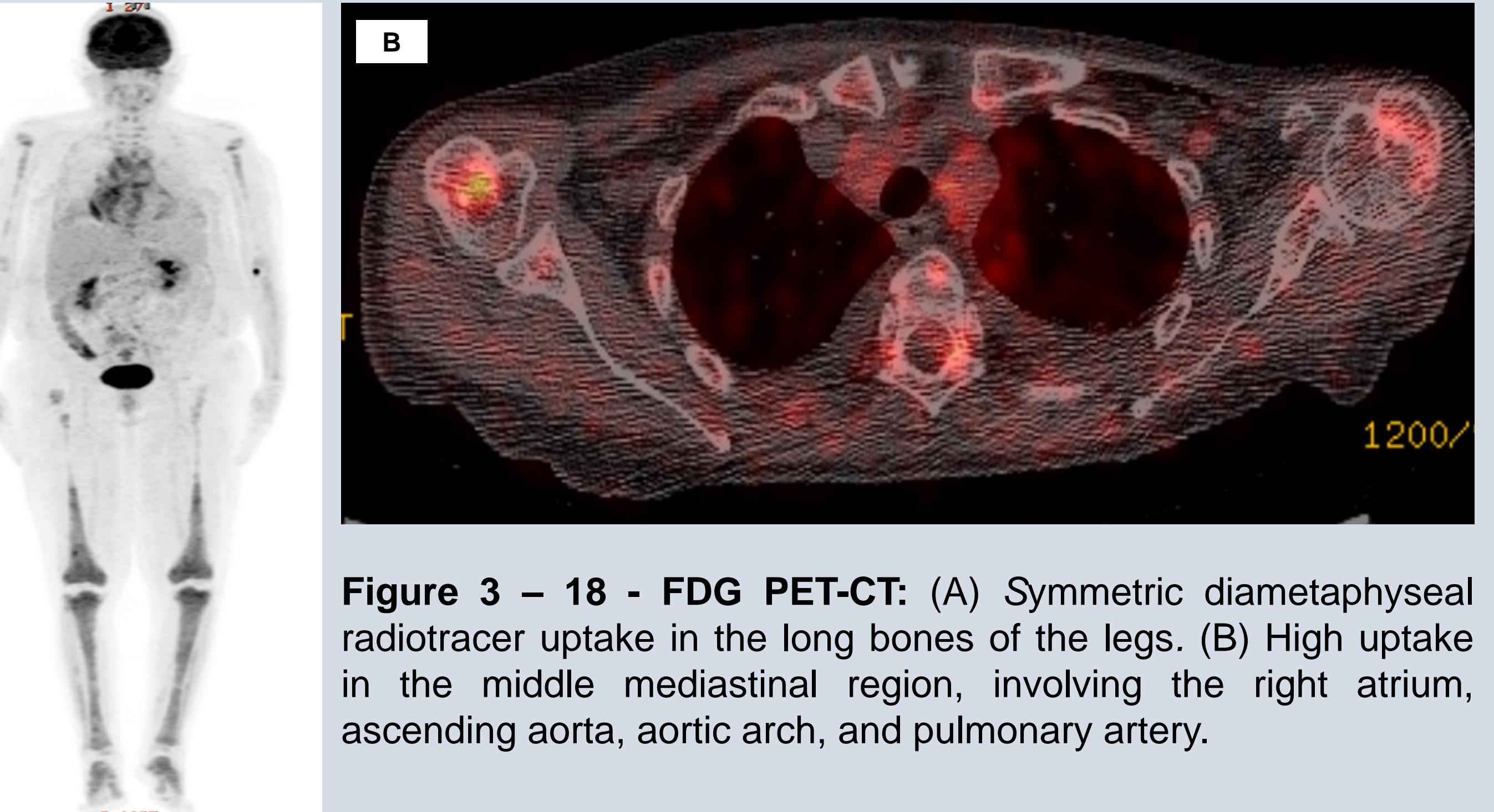


Figure 3 – 18-FDG PET-CT: (A) Symmetric diametaphyseal radiotracer uptake in the long bones of the legs. (B) High uptake in the middle mediastinal region, involving the right atrium, ascending aorta, aortic arch, and pulmonary artery.

## DISCUSSION

- ECD is a rare disease that is difficult to diagnose. Histiocytic disorders are thought to be derived from mononuclear phagocytic cells. The diagnosis is made by histopathological findings.
- The most common abnormality is a circumferential soft-tissue sheathing of the thoracic and abdominal aorta.
- Coronary arterial disease resulting in myocardial infarction has been described.
- Pericardial disease occurs in 40% to 45% of patients and can present with pericarditis, pericardial effusion, and even tamponade.
- Mural pseudo-tumoral infiltration of the RA is present in up to one-third of patients, visualized clearly on MRI as a mass lesion.
- Diffuse infiltration of the myocardium or interatrial septum has been described, occasionally leading to heart failure.
- Treatment involves INF- $\alpha$ , corticosteroids, cytotoxic chemotherapies, serine/threonine kinase inhibitors (vemurafenib and imatinib), radiotherapy, and surgery.

## REFERENCES

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## DISCLOSURES

All the authors have no conflicts of interest to disclose.