

## Erdheim-Chester Disease Presenting with Bradycardia

### Bradikardi ile Prezente Olan Erdheim-Chester Hastalığı

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<sup>1</sup>University of Health Sciences Türkiye, Bursa City Hospital, Clinic of Radiology, Bursa, Türkiye

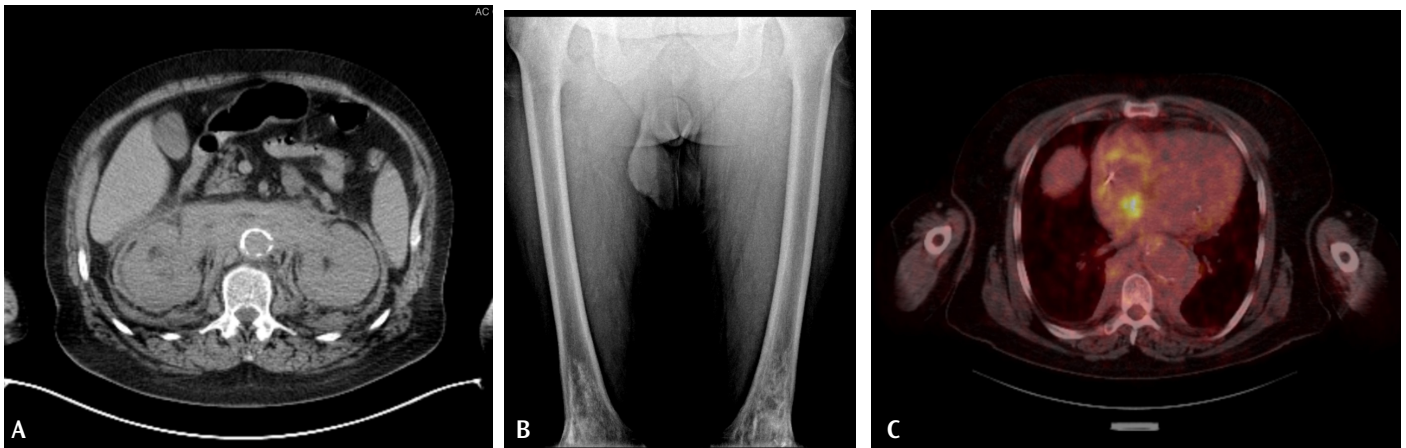
<sup>2</sup>University of Health Sciences Türkiye, Bursa City Hospital, Clinic of Hematology, Bursa, Türkiye

#### To the Editor,

A 65-year-old male patient presented to the emergency department with complaints of headache and nausea. The patient's medical history included hypertension and chronic kidney disease. Electrocardiography revealed bradycardia and a Mobitz type II block. On physical examination, blood pressure was 157/85 mmHg, pulse rate was 43/min, and oxygen saturation was 97% in room air. Blood tests revealed mild leukocytosis with white blood cell count of  $12.62 \times 10^3/\mu\text{L}$  (reference range: 3.9-10.9), creatinine of 1.66 mg/dL (reference range: 0.70-1.20), and increased C-reactive protein of 46.2 mg/L (reference range: 0-5 mg/L). Aminotransferase levels were within the reference ranges. The patient was hospitalized for implantation of a permanent pacemaker. His pulse rate was normalized immediately after the procedure (72/min). During hospitalization, non-contrast abdominal computed tomography performed due to the patient's complaint of back pain revealed para-aortic and perirenal soft tissue densities. The Gerota fascia and Zuckerkandl fascia were thickened and the renal calyces were dilated, indicating possible ureteral involvement (Figure 1A). Lower extremity X-ray showed

osteosclerosis of the distal femurs (Figure 1B). Echocardiography revealed biatrial dilatation and left ventricle hypertrophy. There was mild mitral and tricuspid valve insufficiency. Ejection fraction was 62%. The patient was referred to interventional radiology for tissue diagnosis. Upon examination of the imaging findings before the procedure, biopsy of the perirenal soft tissue was deemed appropriate. The pathology report confirmed CD68<sup>+</sup> and CD1a<sup>-</sup> foamy histiocytes. Further investigation with positron emission tomography revealed a fluorodeoxyglucose-avid pseudotumor on the right atrial wall (Figure 1C).

Erdheim-Chester disease is a non-Langerhans cell histiocytosis characterized by mutation of the *BRAF* gene [1]. It is mostly diagnosed in the 5<sup>th</sup> through 7<sup>th</sup> decades of life. It primarily affects the long bones of the lower extremities but may also involve the retroperitoneum, orbit, paranasal sinus, central nervous system (CNS), lungs, and heart. Diagnosis is challenging due to the diverse presentation and rarity of the disease [2]. Involvement of the CNS presenting with diabetes insipidus, exophthalmos, and cerebellar ataxia is seen in about half of all cases. Computed tomography may reveal the hairy kidney sign, implying perirenal



**Figure 1.** A) The Gerota fascia and Zuckerkandl fascia were thickened and the renal calyces were dilated, indicating possible ureteral involvement. B) Lower extremity X-ray showed osteosclerosis of the distal femurs. C) Positron emission tomography revealed a fluorodeoxyglucose-avid pseudotumor on the right atrial wall.

fat stranding as a typical radiological feature of the disease [3]. Paraaortic soft tissue densities and hydronephrosis secondary to ureteral compression resemble Ormond's disease. Hypertension due to renal artery compression may also be present. Cardiac involvement is not rare in Erdheim-Chester disease, affecting 40% of patients [4]. Cardiac magnetic resonance imaging is the assessment method of choice since it has the best soft tissue resolution. Cardiovascular manifestations include right atrial pseudotumor, high-degree conduction disorders, pericarditis, and coronary artery infiltration [5]. All patients with Erdheim-Chester disease should be investigated for cardiac involvement to ensure appropriate management.

Interferon-alpha has been used extensively in the treatment of Erdheim-Chester disease with response rates of 50%-80%. In recent years, targeted therapies including *BRAF* inhibitors (vemurafenib, encorafenib) have been approved by the U.S. Food and Drug Administration for *BRAF*-V600 mutant Erdheim-Chester disease. Surgery is not curative given the systemic nature of the disease but may be employed in selected cases that require emergent palliation.

**Keywords:** Histiocytosis, Bradycardia, Computed tomography, Erdheim-Chester disease

**Anahtar Sözcükler:** Histiyositoz, Bradikardi, Bilgisayarlı tomografi, Erdheim-Chester hastalığı

## Ethics

**Informed Consent:** Informed consent was obtained from the patient.

## Footnotes

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**Address for Correspondence/Yazışma Adresi:** Fatih Hakan Tufanoğlu, M.D., University of Health Sciences  
Türkiye, Bursa City Hospital, Clinic of Radiology, Bursa, Türkiye  
**E-mail:** fht13@hotmail.com **ORCID:** orcid.org/0000-0002-6720-1937

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