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ADVANCED

CASE REPORT: CLINICAL CASE

Erdheim-Chester Disease

A Case of Right Atrial Involvement and Superior Vena Cava Stenosis

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ABSTRACT

We describe the case of a 79-year-old woman with a history of Erdheim-Chester disease who presented with bradyarrhythmia and infiltration of the superior vena cava and right atrium. This case highlights an important consideration in type of pacemaker placement given the frequency of right atrial involvement in Erdheim-Chester disease. (**Level of Difficulty: Advanced.**) (J Am Coll Cardiol Case Rep 2020;2:1959–65) © 2020 The Authors. Published by Elsevier on behalf of the American College of Cardiology Foundation. This is an open access article under the CC BY-NC-ND license (http://creativecommons.org/licenses/by-nc-nd/4.0/).

HISTORY OF PRESENTATION

A 79-year-old woman was referred from her endocrinologist's office to the hospital for a persistent heart rate in the 30s (beats/min). She has carried a presumed diagnosis of Erdheim-Chester disease (ECD) for 6 years, after incidental imaging demonstrated mass-like infiltration of the right atrium and asymptomatic thickening of the ascending and descending thoracic aorta, right kidney, and right adrenal gland (Figure 1). Invasive work-up with biopsy was recommended at that time; however, the patient deferred further testing and monitoring. On current admission, she denied any chest pain, fatigue, syncope, or presyncope. Her home medications included amlodipine, losartan, metoprolol, furosemide, and rosuvastatin. Physical examination demonstrated a pulse of 38 beats/min, blood pressure of 128/64 mm Hg, respiratory rate of 17 breaths/min, and oxygen saturation of 98% on room air. Lungs were clear to auscultation, and cardiac examination demonstrated bradycardia with regular rhythm, without other significant findings.

LEARNING OBJECTIVES

- To consider use of leadless pacemakers in management of arrhythmias for patients with ECD who have right atrial involvement.
- To understand common structural and electrophysiological cardiac manifestations of ECD.

PAST MEDICAL HISTORY

The patient had a history of hypertension and hyperlipidemia.

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ABBREVIATIONS AND ACRONYMS

ECD = Erdheim-Chester disease

SVC = superior vena cava

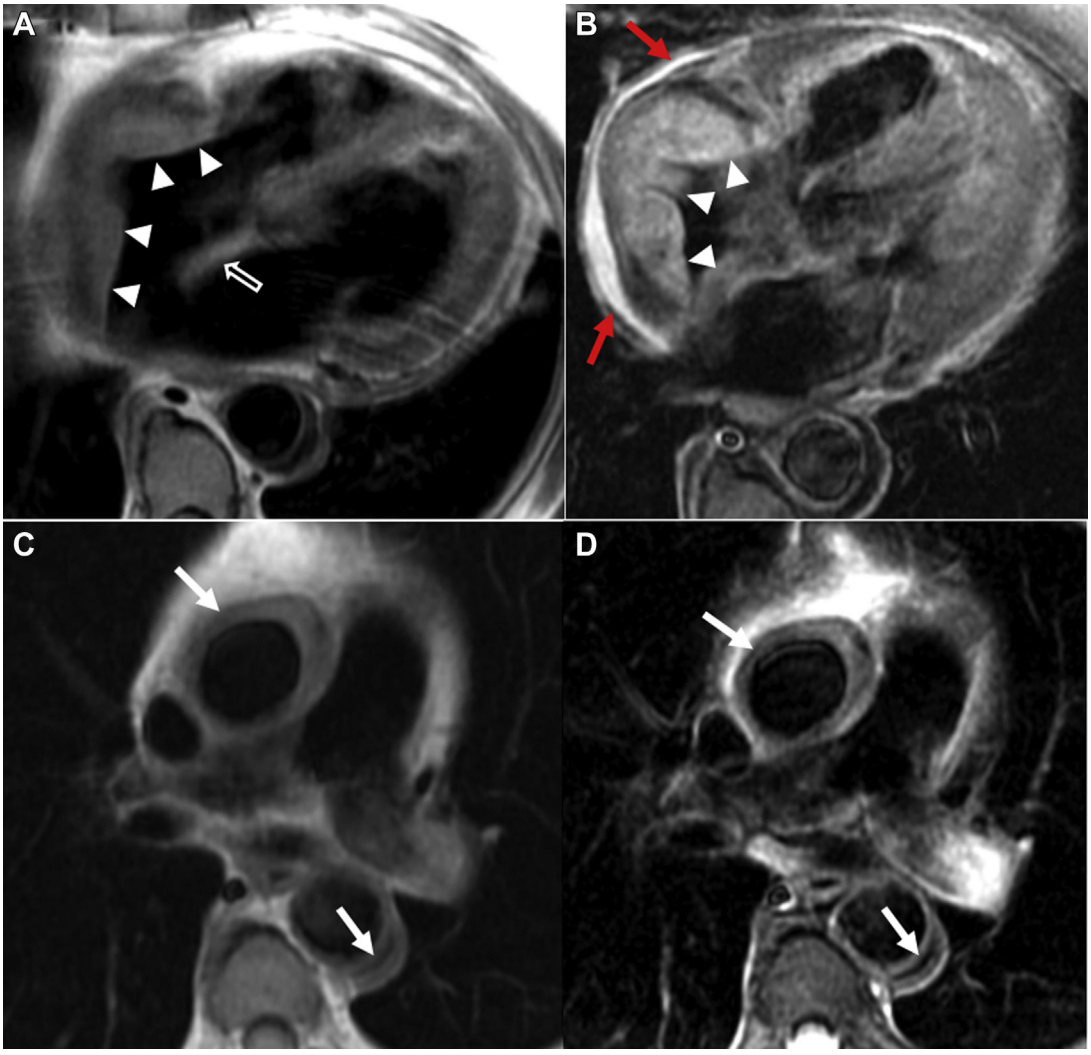
DIFFERENTIAL DIAGNOSIS

The most likely cause of bradycardia in this patient was thought to be progressive infiltration of the conduction system from ECD. Other differential diagnoses included medication-induced bradycardia, myocardial ischemia or infarction, and alternate infiltrate disorders including amyloidosis and lymphoma.

INVESTIGATION

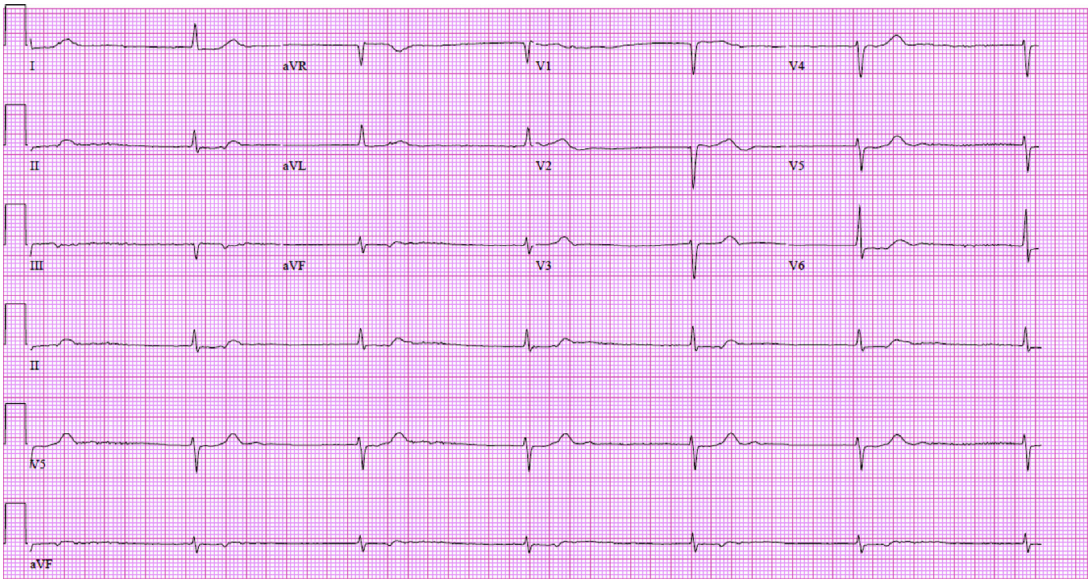
The patient's admission troponin was negative, and the electrocardiogram demonstrated a new junctional escape rhythm (Figure 2). The patient underwent cardiac magnetic resonance imaging that demonstrated infiltrative lesions involving the superior vena cava (SVC), aorta, proximal pulmonary artery, and right atrium (Figures 3A to 3C). There was evidence of

FIGURE 1 Chest Cardiac Magnetic Resonance, 6 Years Earlier



Chest cardiac magnetic resonance demonstrates a mass-like infiltrative lesion involving the right atrium that is most pronounced along the lateral free wall **(A and B, arrowheads),** with milder involvement of the interatrial septum **(A, open arrow)** on **(A)** T_1 and **(B)** T_2 fat-suppressed imaging. A small pericardial effusion is also present **(B, red arrows).** There is periaortic infiltration **(C and D, white arrows)** of the ascending and descending aorta on **(C)** T_1 and **(D)** T_2 fat-suppressed imaging.

FIGURE 2 **Admission Electrocardiogram**



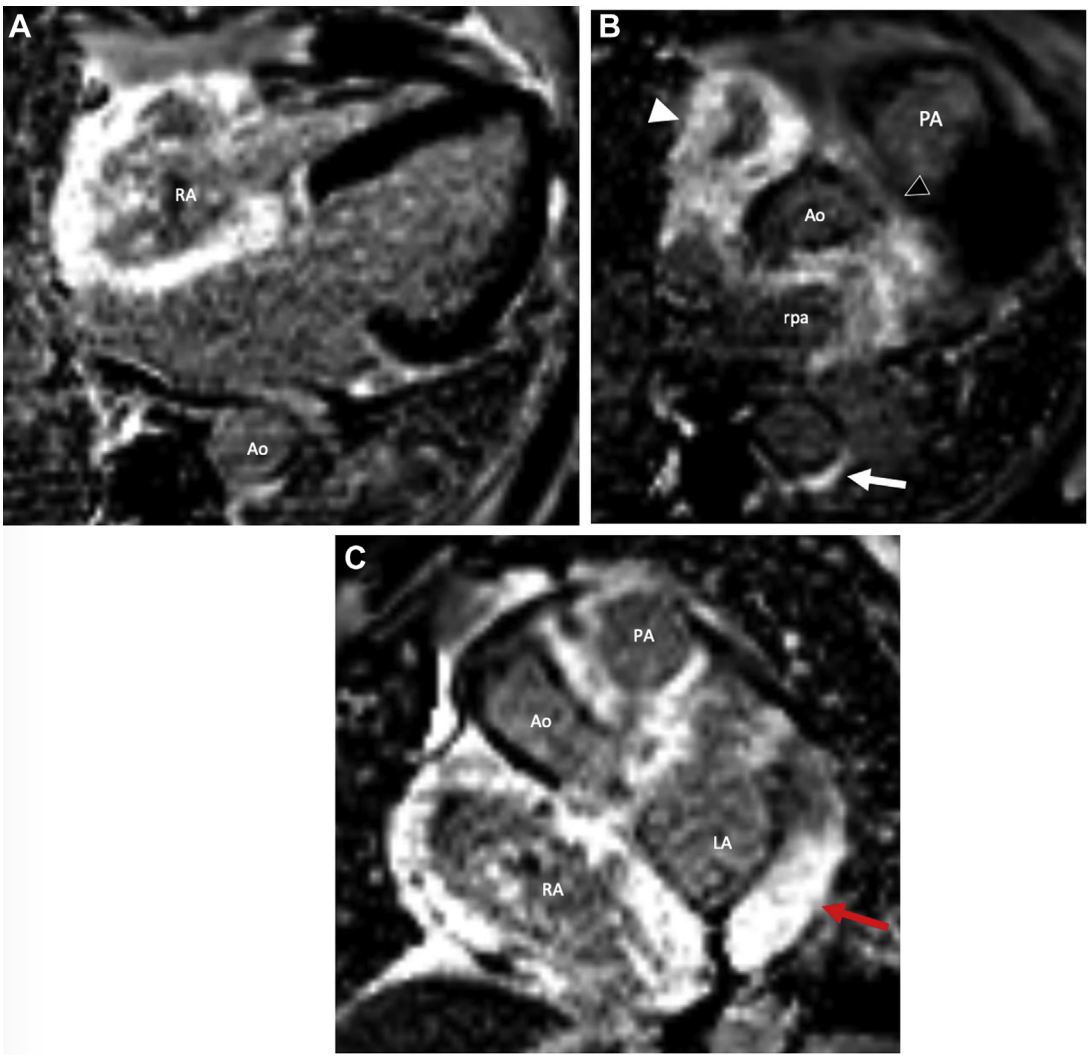
Admission electrocardiogram demonstrated a junctional escape rhythm with narrow QRS complexes and a ventricular rate of 36 beats/min.

significant cardiac disease progression when compared with imaging 6 years earlier (**Figures 4A and 4B**). There was evidence of flow acceleration across the SVC and at the sinoatrial junction, indicating severe stenosis; however, the patient did not clinically present with signs or symptoms of SVC syndrome (**Figures 5A to 5F**). Right-sided heart catheterization was performed through right femoral vein access without an attempt to cross the SVC stenosis. There were minimally elevated pressures, with a right atrial pressure of 10 mm Hg and normal cardiac output and cardiac index. Intracardiac echocardiography-guided endomyocardial biopsy specimens were obtained of the right atrial septum during the procedure, and examination confirmed the diagnosis of ECD, with histological sections demonstrating fibrotic tissue, abundant histiocytes with xanthomatous cytoplasm, and mixed inflammatory cells (**Figure 6**). Immunostaining identified cells with CD163, BRAF V600E, and factor XIIIa positivity and CD1a and S100 negativity, a typical expression pattern in ECD (1).

MANAGEMENT

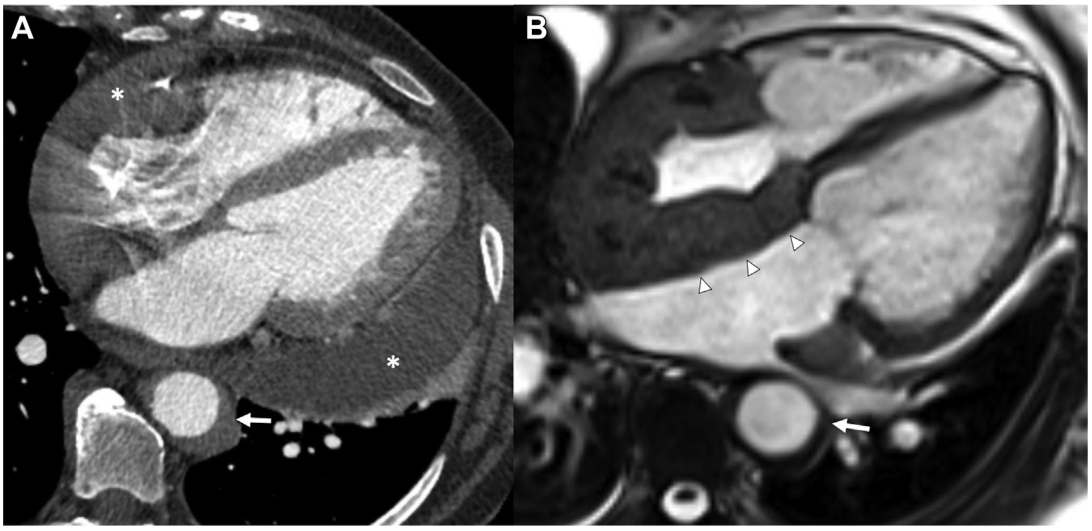
The patient's home metoprolol use was discontinued. After several days of inpatient monitoring, her cardiac rhythm continued to fluctuate among a junctional escape rhythm, ectopic atrial bradycardia, and atrial fibrillation with slow ventricular response. At this point, a decision was made for the patient to undergo pacemaker placement. Given the concern that standard lead placement could worsen SVC stenosis and result in SVC syndrome, a Medtronic Micra leadless pacemaker (Medtronic, Minneapolis, Minnesota) was placed under fluoroscopy through right femoral vein access without complications.

FIGURE 3 Cardiac Magnetic Resonance



Four-chamber phase sensitive inversion recovery images demonstrate **(A)** extensive late gadolinium enhancement of the infiltrative lesion along the right atrium (RA) and atrioventricular sulcus. **(B)** Superior extension of the late gadolinium enhancement, with circumferential encasement of the superior vena cava **(white arrowhead)** and further extension of the infiltrative lesion along the aortic root (Ao), proximal pulmonary artery (PA), and aortopulmonary window **(black arrowhead).** Late gadolinium enhancement is also visible along the descending thoracic aorta **(white arrow). (C)** The structural relationship of the late gadolinium enhancement is better depicted in a short-axis phase sensitive inversion recovery image. Extension of late gadolinium enhancement along the left atrioventricular sulcus is also shown in this image **(red arrow).** LA = left atrium; rpa = right pulmonary artery.

FIGURE 4 Comparison Views of Current Cardiac Magnetic Resonance and Chest Computed Tomography 6 Years Earlier

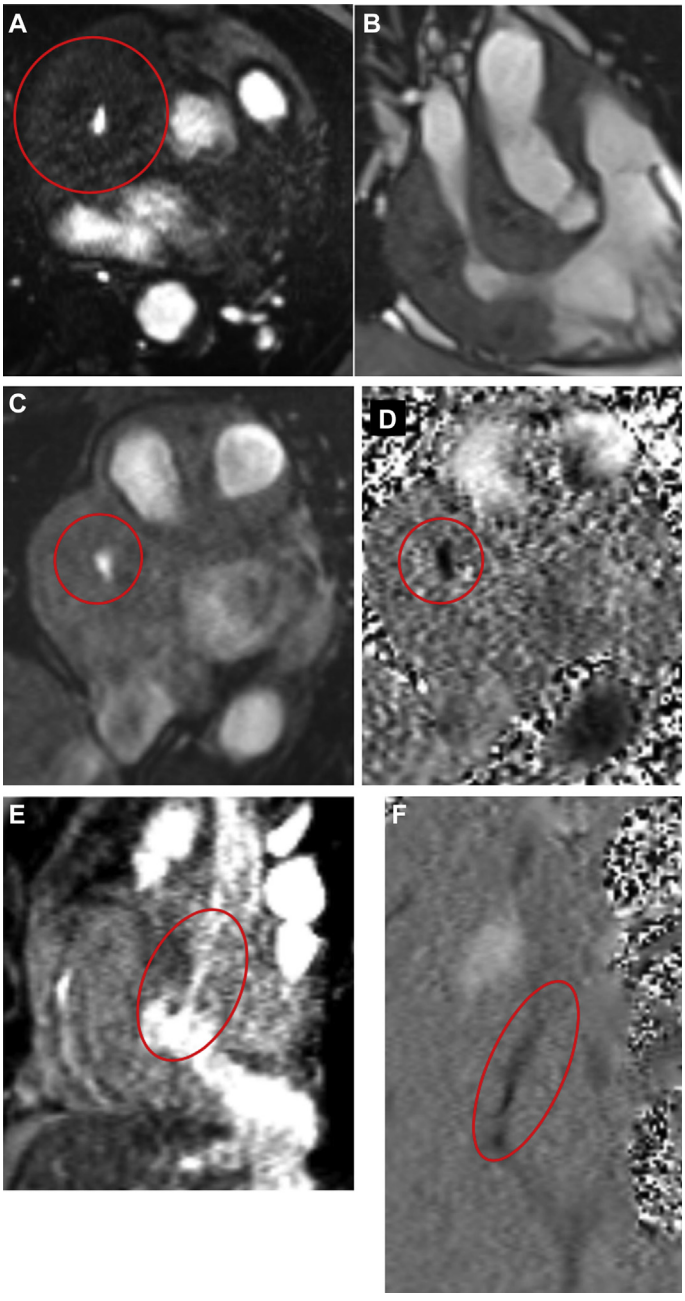


On comparison of **(A)** angiographic chest computed tomography 6 years earlier with **(B)** steady-state free precession cine cardiac magnetic resonance acquisition from current admission, there has been an interval decrease of the patient's pericardial effusion **(asterisk)**, but progression of right atrial infiltration along the interatrial septum **(arrowheads)**. The infiltrative lesion along the descending thoracic aorta has remained **(arrow)**.

DISCUSSION

Although ECD most commonly manifests as non-Langerhans histiocytic infiltration of bones, extraskeletal involvement has been reported to occur in >50% of all cases and is associated with poor clinical prognosis (2,3). Common sites of extraskeletal involvement include the cardiovascular system, retro-orbital tissue, lung, liver, spleen, retroperitoneum, and skin (4). Cardiovascular involvement in ECD has been reported to occur in up to 75% of all patients (4). This most commonly manifests as infiltration of the pericardium and myocardium with a tendency to involve the right atrial myocardium, often with a pseudotumor appearance (5,6). Extensive vascular involvement of the thoracic aorta and SVC has also been reported (7). Typical electrocardiographic abnormalities that have been reported include shortening of the PR interval, sinoatrial block, sinus bradycardia, Q-wave abnormalities, and ST-T wave abnormalities (6). Identifying cardiac involvement is crucial because cardiovascular complications, including hemodynamically significant arrhythmias, cardiomyopathy, myocardial infarction, and severe valvular insufficiency, are frequent causes of death in patients with ECD (2). Cardiac magnetic resonance is an accurate tool for localizing lesions and for assessing the extent of disease involvement (4). This report presents a case of ECD in which cardiac infiltration has extended from the right atrium to involve surrounding vasculature, including the SVC, proximal pulmonary artery, and multiple segments of the aorta. The severity of stenosis at the cavoatrial junction in this case resulted in a decision to place a leadless pacemaker to avoid exacerbating SVC stenosis and inducing SVC syndrome.

FIGURE 5 Evaluation of SVC Stenosis



(A) Selected image from a cine steady-state free precession stacked 4-chamber sequence at the level of the superior cavoatrial junction demonstrates circumferential stenosis of the superior vena cava (SVC) by the infiltrative mass. The degree of SVC stenosis can be best appreciated from a frontal view **(B)**. Magnitude and velocity images from through-plane **(C and D)** and in-plane **(E and F)** phase contrast velocity mapping demonstrate flow acceleration along the stenosed segment of the superior vena cava and superior cavoatrial junction **(red circles)**.

CONCLUSIONS

Although this case illustrates a common presentation of ECD with cardiac involvement, it highlights a unique case in which the severity of SVC stenosis affected decision making in the management of bradyarrhythmia. Given that both conduction abnormalities and right atrial involvement are common cardiac manifestations of ECD, careful consideration should be taken when deciding on the approach and type of pacemaker placement for significant arrhythmias in these patients.

AUTHOR RELATIONSHIP WITH INDUSTRY

The authors have reported that they have no relationships relevant to the contents of this paper to disclose.

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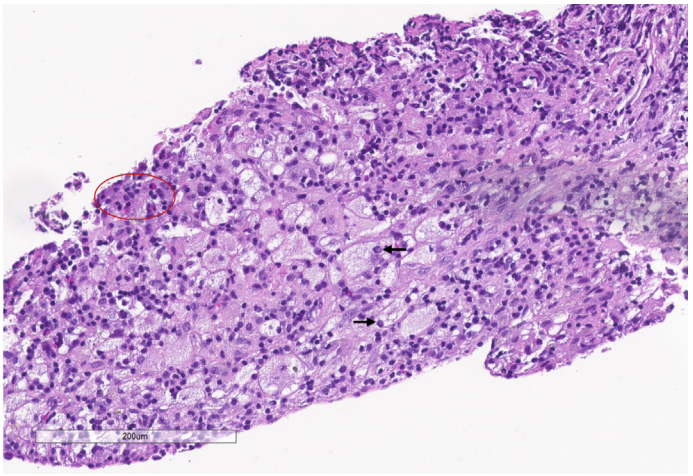
FIGURE 6 Right Atrial Endomyocardial Biopsy



On histological examination, there are relatively abundant histiocytes, many of which demonstrate xanthomatous and vacuolated cytoplasm **(arrows)**. There is also a mild mixed inflammatory infiltrate consisting of plasma cells, lymphocytes, and occasional eosinophils **(circle)**. Fibrotic scarring is also noted in the background.

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KEY WORDS cardiac magnetic resonance, cardiac pacemaker, stenosis