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CASE REPORT

BEGINNER

CLINICAL CASE

Isolated Right-Sided Endomyocardial Fibroelastosis Resulting in Sudden Cardiac Death

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ABSTRACT

Restrictive cardiomyopathy, atrial fibrillation, and cardiac thrombi have been reported with endomyocardial fibroelastosis. This case report examines a unique case of focal endomyocardial fibroelastosis localized to the right ventricle that resulted in sudden cardiac death in an otherwise healthy 21-year-old adult and discusses cardiac magnetic resonance imaging findings in this disease. (**Level of Difficulty: Beginner.**) (J Am Coll Cardiol Case Rep 2019;1:184–7)

HISTORY OF PRESENTATION

A 21-year-old male from Congo presented with 3 episodes of pressure-like chest pain lasting 10 to 15 min over the last 2 months. Episodes were not associated with exertion, including vigorous exercise, and resolved spontaneously. Episodes were not associated with shortness of breath, nausea, or diaphoresis. There were no symptoms of heart failure, including ascites, dyspnea, orthopnea, paroxysmal nocturnal dyspnea, or edema. He never smoked, rarely consumed alcohol, and had no history of drug abuse. He denied any family history of premature coronary artery disease, heart failure, or sudden cardiac death (SCD). The patient had an unremarkable physical examination and had no abnormal heart sounds, murmurs, jugular venous distension, abdominal tenderness or distention, or lower extremity edema.

LEARNING OBJECTIVES

- To identify endomyocardial fibroelastosis as an uncommon etiology of cardiomyopathy and understand appropriate use of various imaging modalities.
- Recognize risk of sudden cardiac death in endomyocardial fibroelastosis.

MEDICAL HISTORY

The patient had malaria at age 13 years.

DIFFERENTIAL DIAGNOSIS

The patient had musculoskeletal chest pain, possible cardiac chest pain of unclear etiology.

INVESTIGATIONS

An electrocardiogram (ECG) showed a first-degree atrioventricular block with a PR interval of 300 ms, high precordial lead voltage, and early repolarization (Figure 1). Transthoracic echocardiography showed right atrial and right ventricular (RV) enlargement and trabeculation from the mid-portion to the apex of the RV with diminished RV systolic function

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(**Figure 2**). Left ventricular size and function were normal. There was no evidence of pulmonary hypertension or valvular disease on the echocardiogram. Cardiac magnetic resonance imaging was obtained for further evaluation, based on his abnormal echo, and it showed enlargement of the proximal to mid-RV and hypertrophy of the distal third (**Figure 3**). Perfusion imaging revealed normal perfusion of apical hypertrophy (**Figure 4**, Video 1), and delayed enhancement imaging did not reveal any abnormal scar or thrombus (**Figure 5**).

MANAGEMENT

Because his chest pain was thought to be unrelated to his cardiac imaging findings, the patient was started on omeprazole and had a noted reduction of chest pain. He had no cardiac complaints for the next 5 years.

FOLLOW-UP

After 5 years, he was found unresponsive and apneic; he was in his usual state of good health the previous day. The autopsy showed right atrial and basal RV dilation, diffuse fibroelastosis of the RV endocardium with mild hypertrophy of the RV wall, and obliteration of the apical RV cavity by fibroelastosis, which are findings consistent with RV endomyocardial fibrosis (EMF).

DISCUSSION

EMF is characterized by fibrosis of the apical to mid-endomyocardium of the RV, left ventricle, or both. It was first described in Uganda in 1948 (1). A high incidence has been reported in Africa, Asia, and South America, and is likely the leading cause of restrictive cardiomyopathy in the developing world. The dilated form is more common and presents as dilated cardiomyopathy with diffuse fibrosis with possible involvement of the papillary muscles and valves (2). The less common contracted form more closely resembles restrictive cardiomyopathy (3). Clinical presentation commonly involves restrictive cardiomyopathy, resulting in right heart failure signs and symptoms (3). Ascites, jugular venous distension, and lower extremity edema are common symptoms. Atrial fibrillation and cardiac thrombi have been reported, with RV involvement and atrial fibrillation implying a poorer prognosis (4). Surgical intervention can improve outcomes to a certain degree (5).

EMF presenting as SCD has not been reported in the literature to the best of our knowledge. Our patient could have developed EMF secondary to malaria infection and the resulting eosinophilia 9 years previously (6). However, he was essentially asymptomatic from a cardiac standpoint. His presentation of chest pressure with abnormal imaging was likely an incidental finding. The involvement of the RV could

ABBREVIATIONS AND ACRONYMS

ECG = electrocardiogram

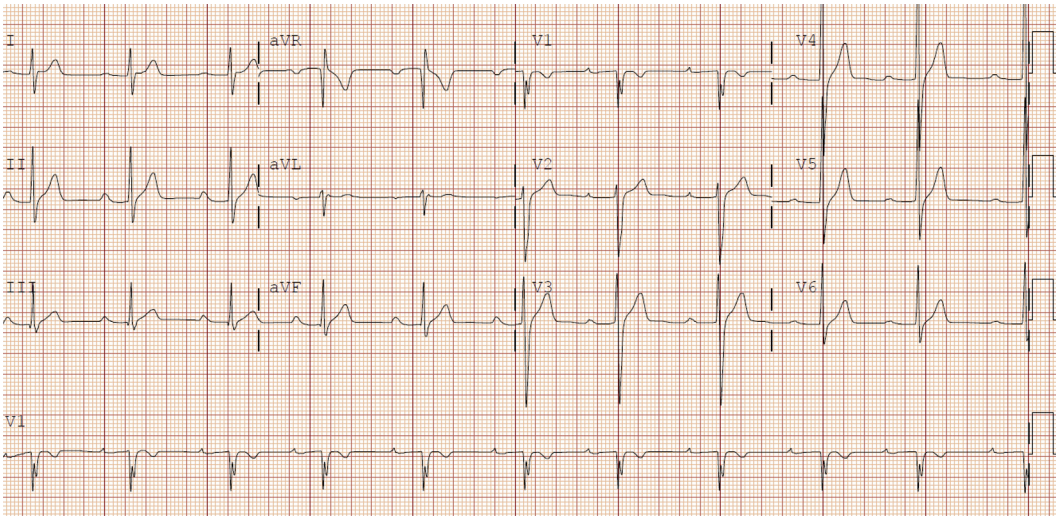
EMF = endomyocardial fibroelastosis

MRI = magnetic resonance imaging

RV = right ventricle

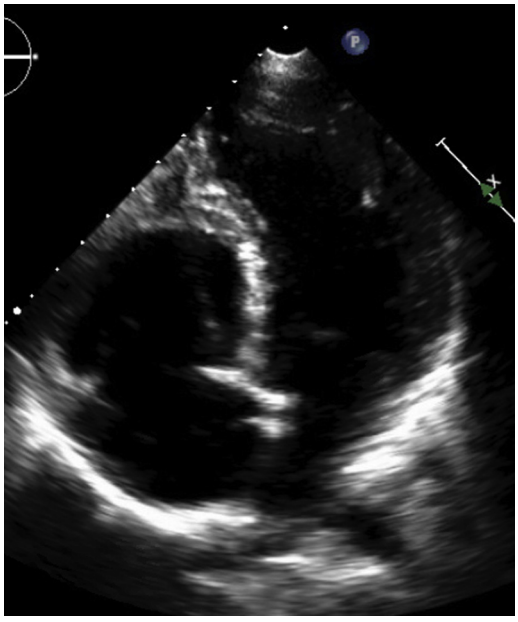
SCD = sudden cardiac death

FIGURE 1 Electrocardiogram With First-Degree Atrioventricular Block



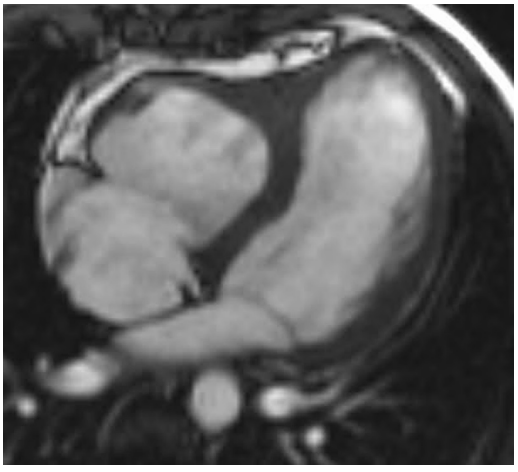
Electrocardiogram showing first-degree atrioventricular block with prolonged PR interval, high precordial lead voltage, and early repolarization.

FIGURE 2 Transthoracic Echocardiogram With Right Atrial and RV Enlargement



Transthoracic echocardiogram with right atrial and right ventricular (RV) enlargement, trabeculation from the midportion to the apex of the RV with diminished RV systolic function.

FIGURE 3 Cardiac Magnetic Resonance Image

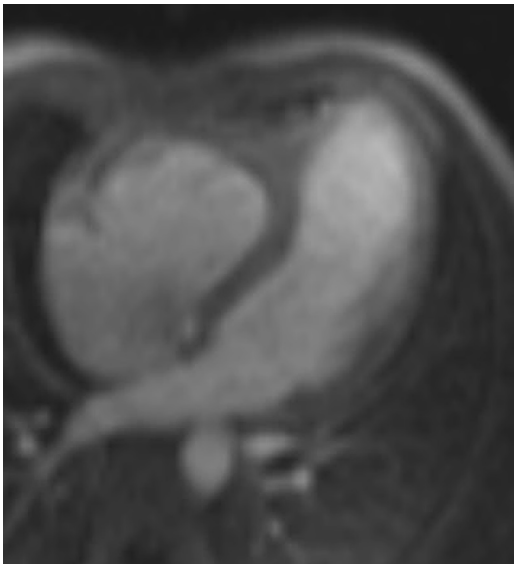


Cardiac magnetic resonance image with enlargement of the proximal to the mid-RV and hypertrophy of the distal third. Abbreviation as in Figure 2.

have given him some predisposition to ventricular arrhythmias that had not been evaluated or described before. Recent data suggested patients with early repolarization were at a higher risk for SCD, especially in patients with structural heart disease (7). It is unclear if this was a contributor to SCD in our patient. A common cause of death in this population is restrictive cardiomyopathy and morbidity due to arrhythmias and ventricular thrombi (3). However, the incidence of EMF could be higher than reported because a large number of patients could be asymptomatic. A large number of patients dying of SCD and arrhythmias in endemic regions could also be due to EMF that resulted in ventricular arrhythmias, which was unknown before.

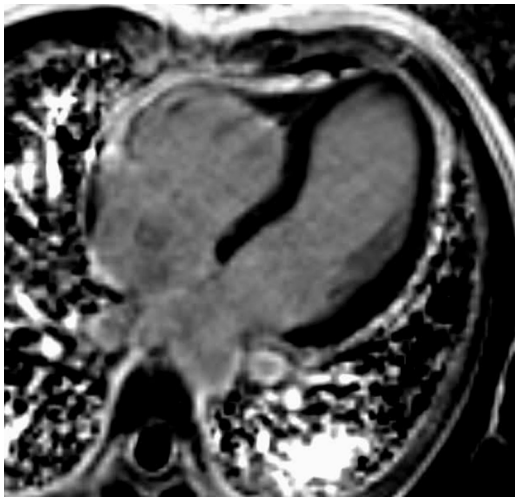
Although tissue diagnosis is the only means for a definitive diagnosis, characteristic imaging findings have been described in the literature that can increase the suspicion for EMF, including late and/or delayed enhancement of endothelium as characteristic findings in patients with EMF (8-10). The tissue deposited in EMF has increased extracellular volume, and thus appears brighter on delayed enhancement imaging because the more vascularized regions wash out the contrast, resulting in the "V pattern" of delayed enhancement (8). However, perfusion imaging and findings on perfusion imaging have not been reported for EMF. Our case demonstrates the uptake of contrast in the entire thickened portion of the myocardium in the RV, which was likely due to increased extracellular volume within the fibrosis. This helps in differentiation among other etiologies,

FIGURE 4 Perfusion Imaging With Normal Perfusion



Perfusion imaging with normal perfusion of the apical hypertrophy. See Video 1.

FIGURE 5 Delayed Enhancement Imaging



Delayed enhancement imaging without any abnormal scar or thrombus.

such as thrombus in the RV apex. We suggest that first-pass perfusion should be included in all cases of suspected EMF.

The cause of death for these patients has been reported as progressively worsening heart failure; however, a case of SCD without evidence of significant heart failure has not been reported in the literature. The fibrotic deposition may certainly predispose these patients to develop ventricular arrhythmias and could be an important and unrecognized cause of SCD in areas of the world where this disease is endemic. This brings up questions about management. Should these patients have additional monitoring for arrhythmias with ambulatory monitoring and should they be considered for implantable cardioverter-defibrillators or life-vests?

CONCLUSIONS

Our case highlights the importance of appropriate diagnosis of EMF with cardiac magnetic resonance imaging and also highlights the risk of SCD in this population. It is unclear if the risk is higher with RV involvement as demonstrated in our patient. Appropriate follow-up, including evaluation for arrhythmias, is important in this patient population.

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KEY WORDS cardiac imaging, cardiac MRI, endomyocardial fibroelastosis, sudden cardiac death

APPENDIX For a supplemental video, please see the online version of this paper.