Circulation: Cardiovascular Imaging

CARDIOVASCULAR IMAGES

Advanced Arrhythmogenic Cardiomyopathy in Former Marathon Runner

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A 60-year-old white female, former nonprofessional agonist marathon runner, diagnosed with arrhythmogenic cardiomyopathy, was admitted to our Emergency Department in 2018 complaining of weakness and palpitations, with ECG evidence of nonsustained ventricular tachycardia.

The patient had been diagnosed in 2009 according to echocardiographic criteria and genetic test, positive for a disease-causing mutation on the plakophilin2 (*PKP2*) gene. She had no living relatives.

Against medical advice, the patient had continued to run and exercise until 2015, when her fitness decreased. In spite of recurrent malignant ventricular arrhythmias and progressive right ventricle (RV) dilatation, she refused both defibrillator implant and heart transplant eligibility assessment and was started on amiodarone.

During hospitalization, cardiac magnetic resonance imaging (1.5-T Avanto; Siemens, Erlangen, Germany) revealed a severely affected, overtrabeculated RV. RV end-diastolic volume was 2.3× greater than normal (248 mL/m^2) and 6.7× greater than the left ventricular end-diastolic volume (RV end-diastolic volume:left ventricular end-diastolic volume=6.7:1). RV walls were thinned (1–2 mm), and dyskinetic areas and diastolic bulging were observed at the inflow tract, outflow tract, and apex, the so-called triangle of dysplasia (Movies 1 and 2 in the Data Supplement). RV systolic function was severely impaired (ejection fraction=18%).

T1-weighted imaging demonstrated diffuse fatty infiltration in the RV wall and trabeculae (Figure 1). Early gadolinium enhancement excluded RV thrombus (Figure 2), whereas transmural late gadolinium enhancement was observed in all the RV segments and trabeculae (Figure 3).

Conversely, left ventricular size, systolic function, and tissue characterization were normal.

Arrhythmogenic cardiomyopathy is an inherited cardiac disease. It is considered a cell adhesion disorder, with many different mutations in genes encoding desmosomal proteins involved in its pathogenesis. Histologically, arrhythmogenic cardiomyopathy is characterized by a progressive replacement of myocardium by fibrofatty tissue,[1] more frequently involving the RV, leading to global dilatation and dysfunction and regional wall motion abnormalities. Physical exercise is a key disease modifier, significantly promoting disease progression toward heart failure, worsening myocytes' mechanical uncoupling, and triggering malignant ventricular arrhythmias.[2] Death is usually arrhythmic or from heart failure; estimated overall mortality is 0.08% to 3.6% per year. Depending on individual circumstances, clinical management consists of exercise restriction, drug therapy, catheter ablation, defibrillators implantation, and in, extreme cases, heart transplant.[3]

The case presented herein illustrates the very end stage of the natural history of arrhythmogenic cardiomyopathy. Not adhering to physical activity restriction, the

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patient sped up the disease progression toward heart failure and favored malignant arrhythmias.

ARTICLE INFORMATION

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Disclosures

None.

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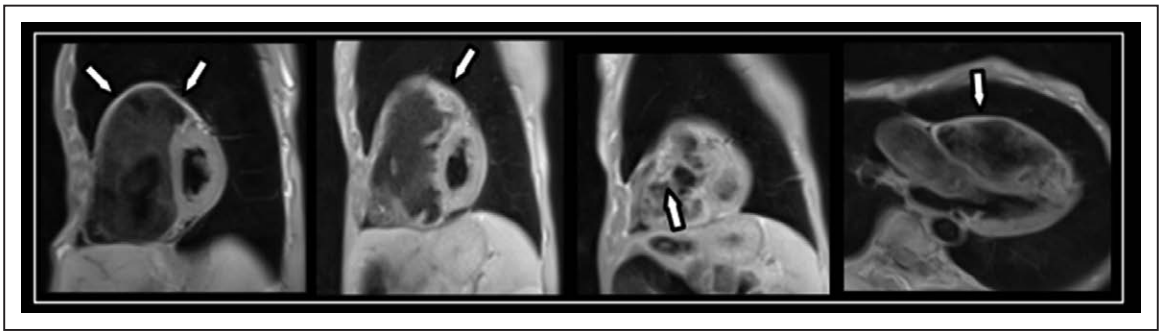

Figure 1. T1-Turbo Spin Echo dark blood images.
From **left** to **right**, basal, midventricular and distal short-axis, and 3-chamber view. Fatty infiltration of the right ventricle can be observed at the outflow tract, free wall, and trabeculae (white arrows).

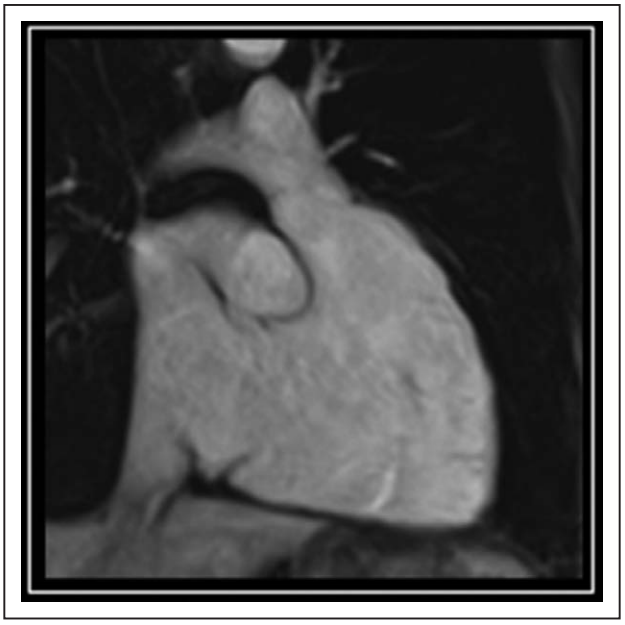

Figure 2. Early gadolinium enhancement excluded thrombus in the right ventricle inflow–outflow view.

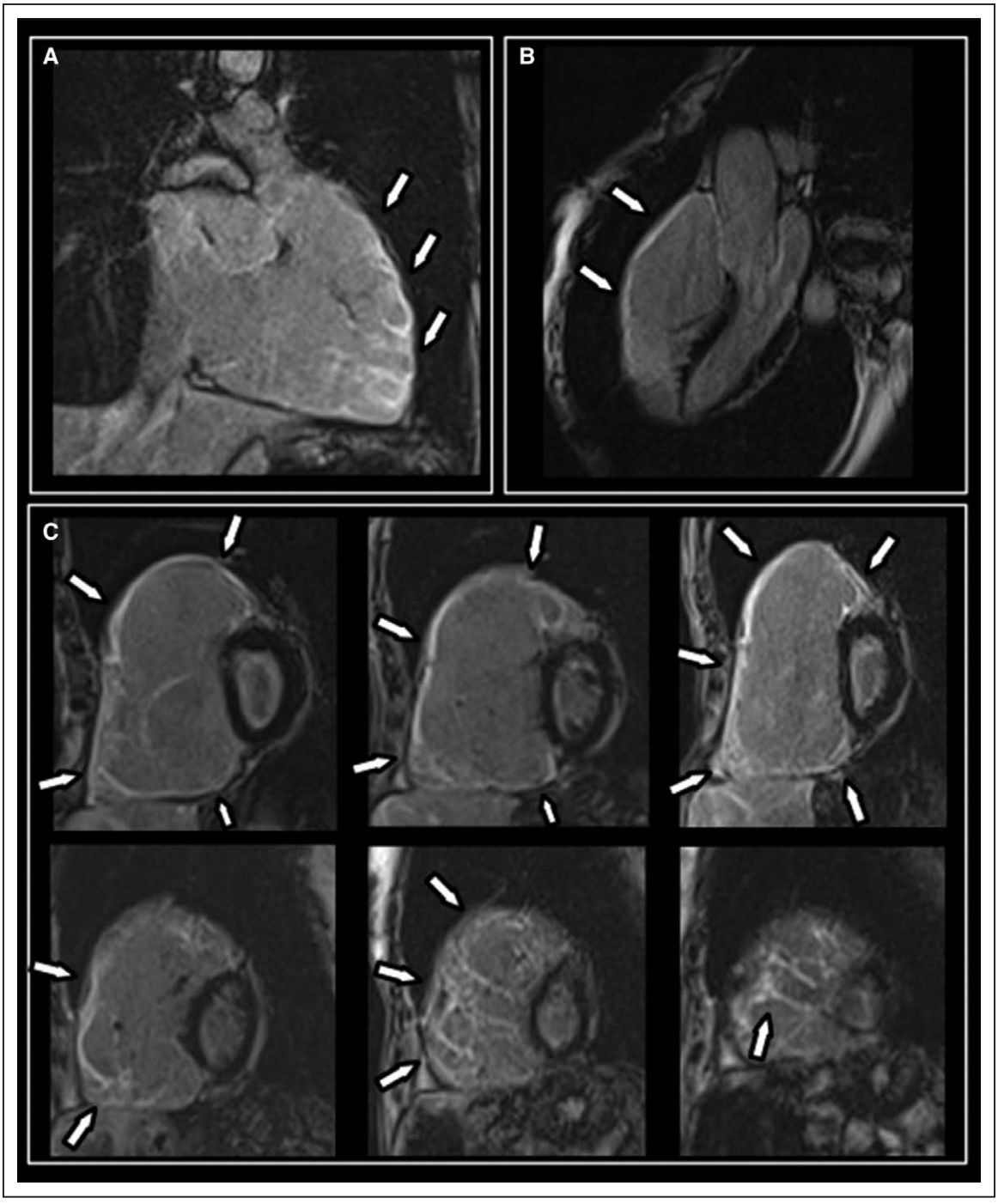

Figure 3. Extensive, transmural late gadolinium enhancement, consistent with fibrous-fatty infiltration, can be observed in all the explored right ventricular walls and trabeculae (white arrows).
A, Right ventricle inflow–outflow view. **B**, Three-chamber view. **C**, Short-axis view.