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CASE REPORT

Recognizing a "Hot Phase" of An Arrhythmogenic Left Ventricular Cardiomyopathy: A Case Report

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Abstract

A 35-year-old male, with a medical history of acute myocarditis, presented with palpitations. Further investigation revealed non-sustained ventricular tachycardia and a slightly reduced left ventricular systolic function. Cardiac magnetic resonance showed extended late gadolinium enhancement of the left ventricle and fat infiltration. Genetic testing was positive for a pathogenic desmoplakin mutation, fulfilling the criteria of arrhythmogenic left ventricular cardiomyopathy.

In conclusion, the authors described a case of a mimicked acute myocarditis at a young age in a patient with an arrhythmogenic left ventricular cardiomyopathy. Therefore, the genetic study is essential for both diagnosis and management.

Keywords: Cardiomyopathies, Desmoplakins, Myocarditis, Cardiac arrhythmias, Sudden death

A 35-year-old male presented to the Cardiology Department following an episode of palpitations. He had no syncope, chest pain, or shortness of breath. He had no cardiovascular risk factors, but a personal history of presumed acute myocarditis at fifteen. No past family history of cardiovascular disease.

In the diagnosis workup, a 24-h ambulatory cardiac monitoring showed frequent polymorphic ventricular ectopic beats and one run of non-sustained ventricular tachycardia (NSVT). The transthoracic echocardiogram revealed a non-dilated left ventricle (LV), with a left ventricular ejection fraction (LVEF) of 53%, associated with slight hypokinesia of the mid-segment of the anterolateral and inferolateral walls, with reduced longitudinal strain (−11 and −14%, respectively, for a normal more than −18%).

Cardiac magnetic resonance (CMR) imaging showed normal-appearing right ventricle (RV); normal LV volumes, with a slightly reduced LVEF (51%); hypokinesia of the mid-segment of the inferior, anterolateral and inferolateral walls. An extended subepicardial circumferential pattern of late gadolinium enhancement of the LV (more than 20% of LV mass − Panel 1A) was noted, and also with the involvement of the right side of interventricular septum. On T2-weighted cine images, a hyperintensity with chemical shift artifact was noted, suggesting fat infiltration.

Genetic testing was positive for a pathogenic heterozygous mutation in the DSP gene [NM_004415.3: c.7000C > T_ p.(Arg2334*)], which encodes desmoplakin (DSP).

Accordingly, the presence of a pathogenic mutation, typical major structural criteria of the LV, and minor RV involvement, fulfil the criteria of arrhythmogenic left ventricular cardiomyopathy (ALVC) [1,2]. Furthermore, DSP gene mutations are associated with a peculiar phenotype characterised by a continuous process of acute myocardial injury, known as "hot phases," which can mimic recurrent episodes of acute myocarditis. In fact, these "hot

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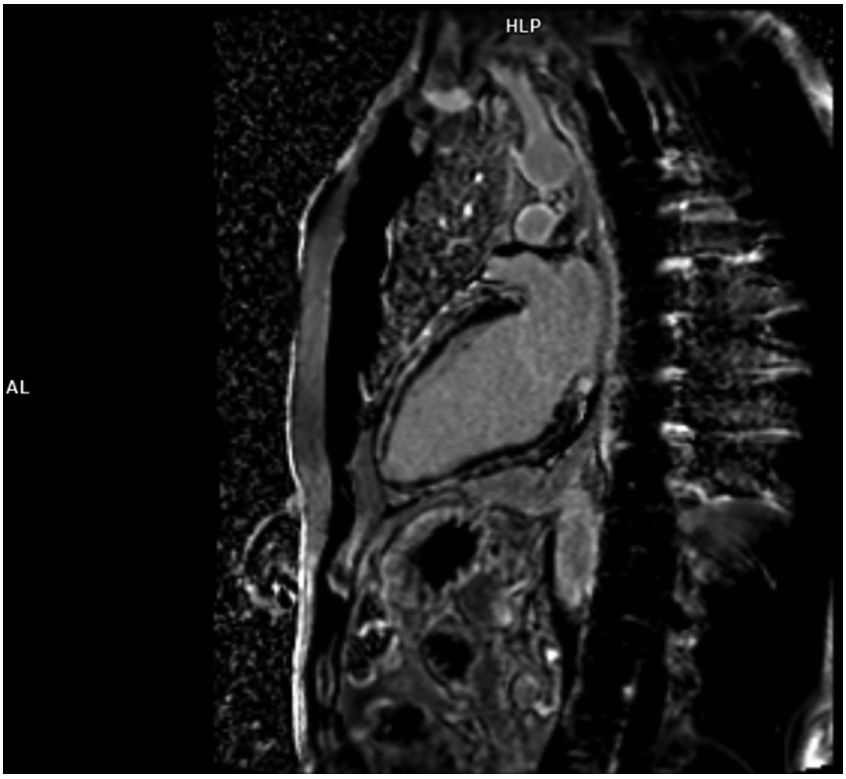
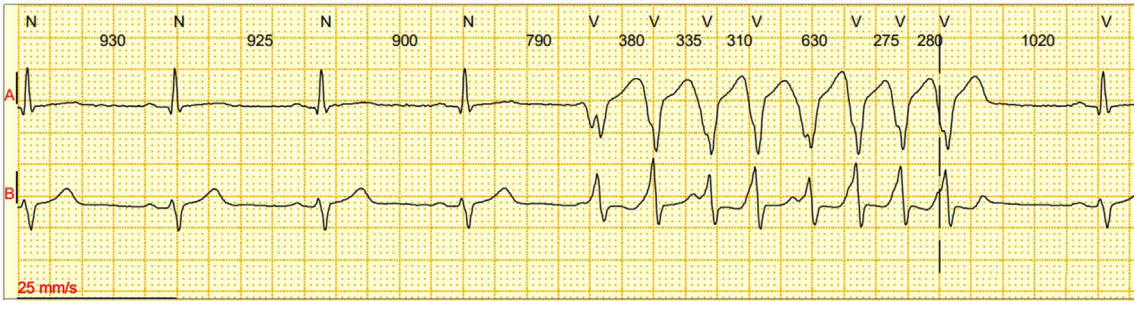


Panel 1A. Extensive late-gadolinium enhancement of the left ventricle.

phases" represent a focal spontaneous necrotic phenomenon of the myocardium followed by an auto-immune response and a complex inflammatory reaction [3]. We raise the possibility that it was what happens at the age of 15. Then, genetic testing for arrhythmogenic cardiomyopathy might be advisable for selected patients with repeated episodes of myocardium inflammation.

Mutations in DSP, the primary force transducer between cardiac desmosomes and intermediate filaments, were first described as a cause of an arrhythmogenic form of predominantly right cardiomyopathy by Rampazzo A et al. and Bauce B. et al. [4,5]. Lately, DSP mutations have also been identified in left-dominant forms and correlated with a high incidence of ventricular arrhythmias, making palpitations the most common symptom at presentation [6–8]. The arrhythmic risk depends on the progressive fibrofatty myocardial replacement, which may present a peculiar circumferential involvement of the LV ("ring pattern") [9].

The current management of ALVC includes exercise restriction, β-blocker therapy, consideration for implantable cardioverter-defibrillator (ICD), and/or catheter ablation. Our patient complies with high-intensity exercise restriction and maximum tolerated dose of β-blocker. During follow-up, a seven-day external cardiac loop recorder detected an increase in the frequency of ventricular ectopic beats (>1000 per 24 h) and an episode of NSVT with eight complexes (Panel 1B). According to current criteria, this patient falls into the intermediate category of SCD



Panel 1B. Nonsustained ventricular tachycardia recorded by the cardiac event recorder.

risk, with an estimated event rate of 1%–10% per year - a strong recommendation for ICD implantation [10–12]. Despite a clear explanation of the risk of sudden death and the importance of ICD as primary prevention, the patient continues to refuse it. Genetic investigation on his son is ongoing, which is essential for early diagnosis of a possible carrier and implanting preventive strategies.

Author contribution

Conception, Visualization: MC, SF. Literature review, Software, Analysis and/or interpretation, Data collection and/or processing: MC, SF, CR. Methodology, Writing - review & editing: MC, SF, CR, HM, JM. Investigation, Resources: MC, SF, CR, HM. Writer-original draft: MC. Supervision: CR, HM, JM.

Conflicts of interest

The authors have no conflicts of interest to declare.

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