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Case Report

Contribution of the Cardiac Magnetic Resonance for the Challenging Diagnosis of Arrhythmogenic Right Ventricular Dysplasia: A Case Report

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Abstract

Arrhythmogenic right ventricular dysplasia (ARVD) is an inherited cardiomyopathy characterized by structural and functional abnormalities in the right ventricle (RV) resulting in ventricular arrhythmias. It is an important cause of sudden cardiac death (SCD) in young adults, accounting for 11% of all cases and 22% of cases among athletes. The structural abnormalities in ARVD result from the fatty infiltration and fibrosis of the RV myocardium. This leads to progressive RV dilatation and dysfunction. The left ventricle (LV) is less commonly involved, and the septum is relatively spared. As the natural history of ARVD is poorly understood, it is difficult to define a definite therapeutic algorithm. The treatment is essentially empiric and based on presentation. We report a case of 22-year-old male presented to the emergency department with recurrent syncope to light workload and, after investigation, cardiac magnetic resonance imaging (CMR) showed a thinning at the midpoint of the anterior RV wall, with dyskinetic movement, and signal hyperintensity after paramagnetic contrast injection, suggesting strongly the diagnosis of ARVD. An implantable cardioverter defibrillator (ICD) was indicated and beta-blocker (atenolol 50 mg twice daily) prescribed. At one year of follow up after ICD, the patient was asymptomatic and did not have any new episodes of syncope during this first year. With this case report, we stress the importance of clinical evaluation in addition to suitable tests with emphasis on CMR to the challenging diagnosis of ARVD.

Keywords: *Arrhythmogenic Right Ventricular Dysplasia; Cardiac Magnetic Resonance Imaging; Right Ventricle; Diagnosis; Implantable Cardioverter Defibrillator*

Abbreviations

ARVD: Arrhythmogenic Right Ventricular Dysplasia; CMR: Cardiac Magnetic Resonance Imaging; ICD: Implantable Cardioverter Defibrillator; LV: Left Ventricle; RV: Right Ventricle; SCD: Sudden Cardiac Death

Introduction

The first description of arrhythmogenic right ventricular dysplasia (ARVD) occurred in 1977 by Fontaine [1]. This is an under recognized clinical condition characterized by ventricular arrhythmias, progressive biventricular dysfunction and sudden cardiac death (SCD). ARVD is characterized macroscopically by replacement of cardiac tissue by fatty/fibrous tissue that initially produces regional abnormalities in the wall motion of the right ventricle (RV), which later produces a biventricular dysfunction. The tissue replacement may also involve areas of the left ventricle (LV) and septum, but generally these are spared [1]. The prevalence in the general population is estimated from 1:1000 to 1:2000 [1]. ARVD has been an important cause of sudden cardiac death (SCD) in young adults, especially in regions such

as northern Italy and Greece, where studies show that ARVD accounts for approximately 11 % of cases in the apparently normal population and 22% in athletes [2,3].

Case Report

A 22-year-old male presented to the emergency department complaining recurrent syncope to light workload (three episodes, with neither prodromes nor traumatic injuries in the last three months). He had a history of smoking for five years and denied family history of sudden death. On the examination, the blood pressure was 132/64 mmHg, the pulse 86 bpm, the respiratory rate 18 breaths per minute, and the oxygen saturation was 95% while breathing in ambient air. The heart sounds were normal and normal vesicular murmur on both lung fields. The electrocardiogram showed sinus rhythm and inverted T wave on leads V1-V4 (Figure 1). The treadmill exercise stress test presented ventricular arrhythmias (isolated ventricular extrasystoles, paired and three episodes of unsustained ventricular tachycardia) on recovery period, with no abnormalities on ST segment. The 24-hour-Holter showed sinus rhythm with mean heart rate of 65 bpm and ventricular extrasystoles polymorphic and non-specific changes in ventricular repolarization. Two-dimensional transthoracic echocardiogram showed structural alterations suggesting ARVD and moderate dysfunction of the RV. Cardiac magnetic resonance imaging (CMR) showed a thinning at the midpoint of the anterior RV wall, with dyskinetic movement, and hyperintensity signal after paramagnetic contrast injection, suggesting strongly the diagnosis of ARVD. Using clinical and radiological criteria, the diagnosis of ARVD was defined. Considering the recurrent episodes of syncope and the findings compatible with ARVD on CMR, an implantable cardioverter defibrillator (ICD) was indicated. A beta-blocker (atenolol 50 mg twice daily) was initiated. The patient was followed up in our outpatient clinic and one year after the implantation of the cardioverter defibrillator, the patient has not presented new episodes of syncope.

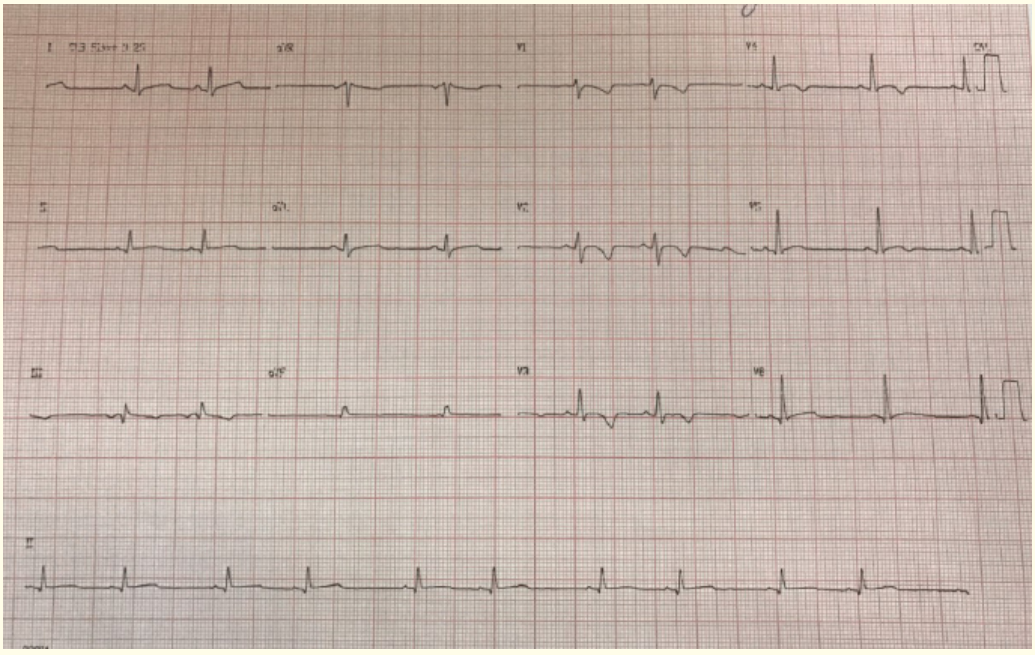
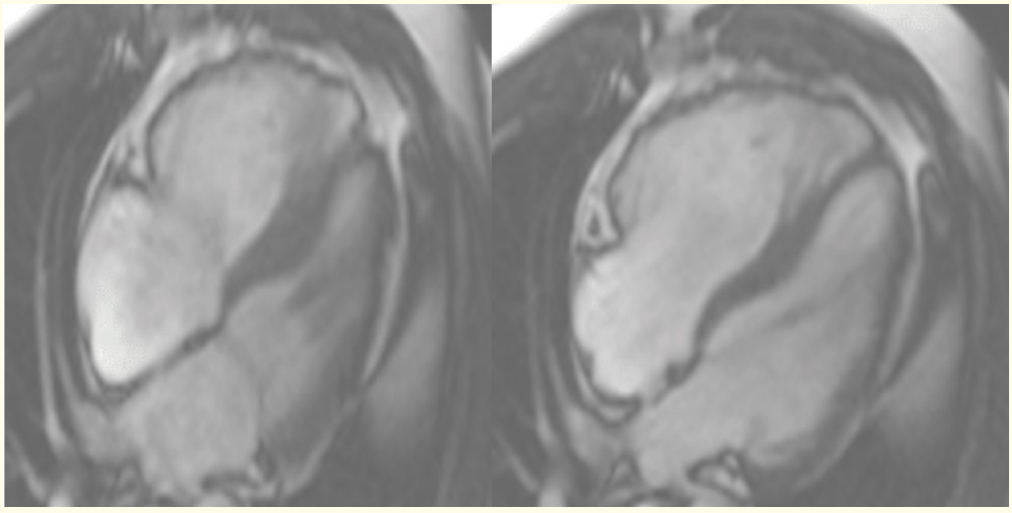


Figure 1: *Electrocardiogram showing sinus rhythm and inverted T wave on leads V1-V4.*



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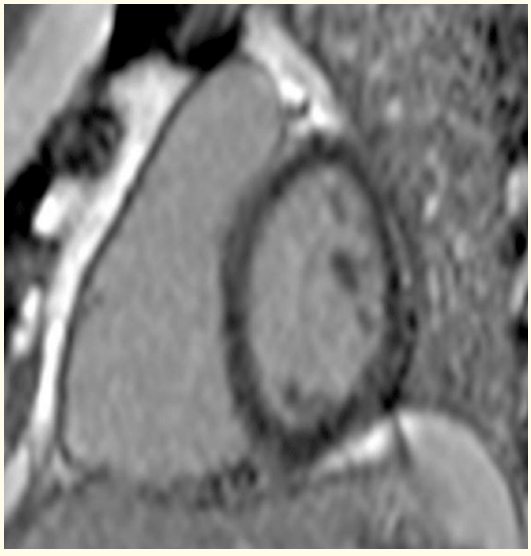

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Figure 2: *Functional analysis shows global hypokinesia of the right ventricle (RV) free wall; A: steady-state free precession (SSFP) multi shot sequence 4-chamber view at end-systole; B: SSFP multi shot sequence 4-chamber view at end-diastole; C: delayed enhancement short axis.*

Discussion

The diagnosis of ARVD can be challenging, requiring a high degree of clinical suspicion and often multiple diagnostic tests or procedures to the proper diagnosis. The most common location of greasy deposition is between the anterior infundibulum, ventricular apex and the diaphragmatic portion of the RV, known as the "triangle of dysplasia". Patients with ARVD may present symptoms at any age, being

more common in youth (33 ± 14 years), with predominance of males (3:1 ratio) [4]. Several genes and locus are associated with ARVD, and autosomal recessive and dominant forms of inheritance are described. Many, but not all, genes code for the desmosomal proteins. The genes involved include desmoplaquine, junction plakoglobin, cardiac ryanodine receptor-2 and transforming growth factor B3. These genes have prognostic importance in ARVD [4].

Many of the clinical findings and additional routine tests have low sensitivity and specificity for ARVD. Thus, diagnostic criteria have been published in an effort to standardize the process of the correct diagnosis [5]. In 2010 a task force was carried out, with the objective to define the diagnostic criteria for ARVD. The major criteria include the demonstration of severe abnormalities in ventricular wall motion, replacement by fibrous tissue in the biopsy-proven endomyocardium, epsilon waves, and the histologically confirmed family history of the disease. Minor criteria include mild changes in ventricular function, inverted T wave in right precordial leads, late potential on high resolution electrocardiogram, ventricular tachycardia with frequent right ventricular morphology or premature ventricular complex (> 1000 in 24 hours monitoring). The diagnosis has been established with two major criteria, or one major and two minor, or four minor criteria [6]. The gold standard for the diagnosis of ARVD remains the endomyocardial biopsy, but even the latter has a relatively low sensitivity to make the diagnosis, since the biopsy site occurs in the wall of the septum, a site that is not very commonly affected by the disease [5].

Beta-blocker treatment is recommended in all patients with ARVD. Antiarrhythmic drugs can reduce the frequency and suppress the ability of induction of sustained and unsustained ventricular arrhythmias in patients with ARVD [9]. However, antiarrhythmic drugs have not been shown to reduce the risk of sudden cardiac death in the disease [7]. Radiofrequency ablation (RFA) is not indicated as a primary or exclusive therapy for the treatment of ventricular arrhythmias in patients with ARVD. RFA can successfully treat some of the arrhythmogenic foci, but due to the heterogeneity and progressive nature of this disease, RFA is not a definitive therapy [8].

Patients with ARVD should be prevented from competitive activities and high intensity recreation activities. Recreational activities of moderate intensity should be approached with caution [7]. ICD is appropriate for the secondary prevention of MSC in people who have sustained ventricular arrhythmia. It is also considered appropriate for primary prevention from SCD in selected patients considered to be at high risk: extensive RV disease, involvement of the LV, or syncope consistent with a documented ventricular tachyarrhythmia [8]. Due to the low prevalence of ARVD and the lack of randomized clinical trials, precise indications for ICD for primary prevention of SCD have not been developed, and recommendations based on societies guidelines are primarily based on consensus of the experts [8].

Although patients with ARVD have a risk of developing heart failure and ventricular arrhythmias, most patients with ICD and suitable pharmacological treatment have good clinical outcomes. In a meta-analysis of 610 patients using ICD and drug treatment, the annual rate of cardiac death, non-cardiac death and need for heart transplantation were 0.9%, 0.8%, and 0.9%, respectively [10].

In this case report, the patient presented recurrent syncope, T wave inversion in the right precordial leads, and RV wall motion abnormalities in the two-dimensional transthoracic echocardiography in addition to the CMR imaging consistent with diagnosis of ARVD. The patient received an ICD associated with drug therapy. After eight months of follow-up, the patient was asymptomatic, sinus rhythm on electrocardiogram and without any new episodes of syncope.

Conclusion

This case shows that an applicable clinical evaluation in addition to suitable tests with emphasis on CMR contribute to the challenging diagnosis of ARVD. ICD and drug therapy are important components of the appropriate treatment.

Conflict of Interest

The authors declare that there are no financial interest or any conflict of interest.

Bibliography

1. European Heart Rhythm Association, Heart Rhythm Society., *et al*. "ACC/AHA/ESC 2006 guidelines for management of patients with ventricular arrhythmias and the prevention of sudden cardiac death: a report of the American College of Cardiology/American Heart Association Task Force and the European Society of Cardiology Committee for Practice Guidelines (Writing Committee to Develop Guidelines for Management of Patients With Ventricular Arrhythmias and the Prevention of Sudden Cardiac Death)". *Journal of the American College of Cardiology* 48.5 (2006): e247-e346.

2. Te Riele AS., *et al*. "Mutation-positive arrhythmogenic right ventricular dysplasia/cardiomyopathy: the triangle of dysplasia displaced". *Journal of Cardiovascular Electrophysiology* 24.12 (2013): 1311-1320.

3. Gemayel C., *et al*. "Arrhythmogenic right ventricular cardiomyopathy". *Journal of the American College of Cardiology* 38.7 (2001): 1773-1781.

4. Richardson P., *et al*. "Report of the 1995 World Health Organization/International Society and Federation of Cardiology Task Force on the Definition and Classification of cardiomyopathies". *Circulation* 93.5 (1996): 841-842.

5. Anderson EL. "Arrhythmogenic right ventricular dysplasia". *American Family Physician* 73.8 (2006): 1391-1398.

6. Serrano Jr CV. "Tratado de cardiologia SOCESP". 2nd edition. São Paulo: Manole (2009).

7. Marcus FI., *et al*. "Diagnosis of arrhythmogenic right ventricular cardiomyopathy/dysplasia: proposed modification of the task force criteria". *Circulation* 121 (2010): 1533-1541.

8. Maron BJ., *et al*. "Task Force 4: HCM and other cardiomyopathies, mitral valve prolapse, myocarditis, and Marfan syndrome". *Journal of the American College of Cardiology* 45.8 (2005): 1340-1345.

9. Corrado D., *et al*. "Treatment of arrhythmogenic right ventricular cardiomyopathy/dysplasia: an international task force consensus statement". *European Heart Journal* 36.46 (2015): 3227-3237.

10. Schinkel AF. "Implantable cardioverter defibrillators in arrhythmogenic right ventricular dysplasia/cardiomyopathy: patient outcomes, incidence of appropriate and inappropriate interventions, and complications". *Circulation: Arrhythmia and Electrophysiology* 6.3 (2013): 562-568.

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