Introduction

Arrhythmogenic right ventricular dysplasia (ARVD) is a rare, inherited disease that predominantly affects the right ventricle (RV) and predisposes to ventricular arrhythmias and increased risk of sudden death.¹ The estimated prevalence of the disease in the general population is of 1 in 1000 to 1 in 5000 persons.¹ It is accountable for 3% to 10% of the sudden deaths reported in patients under 65 years.²

Originally, it was described as a heart disease that affects only the right ventricular. However, it is currently known that there are clinical and pathological evidences that the left ventricular (LV) may also be affected.¹ The impairment of the LF depends on the present genetic mutation and may occur in the early stages of the disease.¹

The main pathology of the ARVD is the myocyte loss with fibrofatty replacement.¹ Genetic defects in proteins from the desmosomal complex may impair the mechanical coupling among individual cells, leading to the uncoupling of myocytes, particularly in conditions that increase the strain in the myocardium.¹ As a consequence, there is a disruption in the junctions and adhesions between such cells, leading to inflammation, fibrosis and myocardium replacement for fat deposit¹. This substrate results in the development of a complex heterogeneous scar, which increases the susceptibility to ventricular arrhythmia.¹⁻⁴

Patients usually present early signs and symptoms of the disease in the 2nd to 5th decade of life, which are mainly palpitations, vertigo, syncope or sudden death.¹

Case report

Male patient, 45 years old, married, fisherman, from Itaboraí-RJ. He was admitted to the emergency of the university hospital due to asymptomatic sinus bradycardia, presented during monitoring for elective performance of myocardial scintigraphy recommended by his cardiologist. He was under regular use of amiodarone 400 mg/day for 5 months. He was diagnosed with sinus bradycardia by amiodarone impregnation. Then, the heart rate returned to normal levels following suspension of amiodarone and use of atropine. Three days later, the patient presented a frame of tachyarrhythmia associated with lipohyemia, with electrocardiogram showing tachycardia with wide QRS, compatible with ventricular tachycardia (Figure 1).

The patient was being investigated by his cardiologist for 2 years, due to syncopal episodes. In one of the episodes, an ECG was performed, which evidenced tachycardia with wide QRS, requiring electric cardioversion. After this event, the patient initiated regular use of amiodarone 400 mg/day. He had no history of sudden death in the family.

He presented exams made in the previous year, on an outpatient basis, by his cardiologist, such as ergometric test, cardiac catheterization, transthoracic echocardiography and cineangiocardiography without expressive alterations. He also presented 24-hour Holter with sinus rhythm, average heart rate of 57 beats per minute, presence of supraventricular and ventricular extrasystoles (1065); one episode of non-sustained ventricular tachycardia (3 complexes) and one episode of non-sustained paroxysmal supraventricular tachycardia.
Ventricular tachycardia occurred after the suspension of the amiodarone due to sinus bradycardia.

The admission electrocardiogram evidenced sinus rhythm, with T-wave inversion in the anterior and inferior ventricular wall (Figure 2). The admission echocardiogram showed relaxation deficit, mild mitral and aortic insufficiency and mild increase on the LF.

The patient underwent the performance of cardiac magnetic resonance imaging (CMRI), which evidenced the right atrium at the upper limit of normality, increased RV, with areas of dyskinesia and aneurysmal formation in the intersection of the free wall with the inferior of the
RV, measuring approximately 3.7 x 1.4 x 1.3 cm. The index of the final RV diastolic volume was calculated at 97.9 ml/m². Important global systolic dysfunction with ejection fraction of the RV estimated at 29%. He also presented signals of fatty infiltration in the free and inferior RV wall and in the LV interventricular septum and apical lateral wall (Figure 3).

The left cardiac chambers are normal-sized, and the global systolic function is preserved.

The patient underwent an implantable cardiac defibrillator (ICD implant) uneventfully. He was kept with amiodarone 400 mg/day in association with the ICD and, so far, evolves uneventfully.
Figure 3
Cardiac Magnetic Resonance in the long axis, four chambers, and e B and Cardiac MR in the short axis, C and D.
(A): cine-MR sequence with clear blood, showing the aneurysm and dyskinesia areas of the right ventricle (arrowheads), in addition to chemical shift artifact in the interventricular septum (arrow), typical of fat infiltration and the large fatty infiltration in the lateral wall of the left ventricular, with thinning of the muscle in the same segment (star). (B): anatomical sequence and of tissue characterization known as delayed enhancement. We managed to identify the fat and the absence of fibrosis in the intact muscles of the left ventricular. (C): anatomic sequence and of tissue characterization with double inversion and recovery pulse, with dark blood, showing the fat tissue infiltrating (arrowheads) the free and inferior wall of the right ventricular, the inferior and lateral wall of the left ventricular and the interventricular septum (arrow). (D): anatomic sequence and of tissue characterization known as delayed enhancement. We managed to identify the fat and the absence of fibrosis in the intact muscles of the left ventricular.

Discussion

In 1994 and 2010, an International Task Force (ITF) document proposed guidelines for the standardized diagnosis of ARVD, which relies on a score system with major and minor criteria, based on the demonstration of the combination of defects in the RV morphology and function, electrocardiographic abnormalities, ventricular arrhythmias, family history, results of genetic testing and tissue analysis.1,3,4

Inverted T-wave in right precordial leads (v1-v3) is a very common manifestation in the ECG of the disease.1,3 The presence of ventricular extrasystoles or sustained or non-sustained ventricular tachycardia evidenced by the 24-hour Holter are consistent with the ARVD diagnosis.1,3

The findings that suggest the ARVD by the echocardiogram include abnormal global or targeted movement of the RV wall, cavitary dilation with systolic dysfunction and hypertrophic RV.3 The dilatation of the RV outflow tract (diameter greater than 30 mm) was reported as having the highest sensibility and specificity of the echocardiographic parameters for diagnosis of ARVD1.

The CMRI with a fast and objective protocol brings the benefit of identifying fat and myocardial fibrosis, as well as assessing the global or targeted structure and function of the RV and of the LV.1,3 Currently, the CMRI is the exam of choice in tissue characterization and, mainly, in the detection of akinesia, dyskinesia or dyssynchrony of the RV 1. These findings, associated to the ejection fraction of the RV < 40% or diastolic volume index of the enlarged RV, > 110 mL/m² for men and > 100 mL/m² for women, are considered the major criteria by the method.1,3
The Cardiac Computed Tomography (CCT) is also capable to identify fat infiltration in the wall of the RV and of the LV or areas of alteration of segmental contractility, as well as quantify the global function. The CCT has been admittedly used for correlation with electrophysiology maps for demonstration of the arrhythmia foci in ARVD. We must stress that, in the monitoring of patients with ICD, the CCT may be used for quantification of the final diastolic volume and evaluation of the progress of the disease.

The immunohistochemical analysis by means of biopsy for ARVD diagnosis is little used currently; however, it is still important, because only it provides the definitive diagnosis.

Although the genetic tests may be used for screening diagnosis, its practical use is hindered due to the presence of very variable mutations and for having a very variable, indeterminate genotype / phenotype relationship. Therefore, the tracking of relatives is still made by the electrocardiogram, echocardiogram and 24-h Holter. Genetic tests, however, are useful in risk stratification, as certain mutations are related to worse prognosis. In the case at hand, the patient was oriented in the hospital discharge to inform his first-degree relatives to attend the hospital for ARVD screening.

The handling of patients with ARVD consists of the risk stratification and decision on ICD, relief of symptoms and prevention of shocks by the ICD, catheter ablation of ventricular arrhythmias and restriction of physical exercises. The ICD is indicated for patients with history of sudden death, sustained ventricular tachycardia, syncope of arrhythmic origin or high degree of ventricular ectopy or non-sustained ventricular tachycardia. Pharmacological therapy and prevention of shocks can be made with beta-blockers, sotalol or amiodarone.

Although it is not always remembered, the involvement of the LV is not rare. There are mutations that are more related to the LV involvement, with their own characteristics. According to revision by Berte et al., in a total of 32 patients with ARVD, fibrosis and abnormal movements of the walls of the LV in RNM were found in MRI in 14 (64%) and 2 (9%) patients, respectively, while fat was found in the computed tomography in 21 (66%) patients. These results reinforce the necessity for evaluation of alternative strategies to identify dysplasia in the LV. The importance of the involvement of the LV lies in the fact that it may contribute with the arrhythmogenicity of the disease.

Author contributions

Acquisition of data: Reetz KB, Nacif M, Silva EN. Writing of the manuscript: Reetz KB, Villacorta Junior H. Critical revision of the manuscript for intellectual content: Nacif M, Silva EN, Martins W, Villacorta Junior H.

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References