Arrythmogenic Right Ventricular Dysplasia

Rogério Ferreira da Silva[1], Karina Morgarbel[1], Christian Moreno Luize[1], Carla Gonçalves Rosa[1], Marcelo Romano[2], Ieda Maria Liguori[1]

Hospital do Coração (HCOR) - Associação do Sanatório Sírio, São Paulo, SP[1]; Universidade Oeste Paulista (UNOESTE), Presidente Prudente, SP[2] - Brazil.

Arritmogenic right ventricular dysplasia (ARVD) is characterized by the gradual replacement of myocytes by adipose and fibrous tissue. Described in 1977, is considered a potentially lethal cause of cardiac disease poorly understood. This disorder usually involves the right ventricle and has been associated with arrthymia, heart failure, and sudden death. In this paper, we report a case of a 25-years-old patient with syncope associated with ventricular extrasystoles. A magnetic resonance imaging was performed and showed findings that support ARVD diagnose.

Introduction

ARVD – described in 1977 by Fontaine et al[1] – is a potentially lethal heart disease not yet fully understood[2,3]. It affects primarily the right ventricle by fibrofatty replacement of the RV, and has been associated to arrhythmia, heart failure and sudden death[3-5].

Estimated prevalence is 1:5000, although diagnosis does pose difficulties[3]. It accounts for 3% to 4% of deaths among sport practitioners, and 5% of sudden deaths before 65 years of age[2].

Family predisposition was described in 1982 by Marcus et al.[6] Approximately 30% of diagnosed patients refer a family history[2,3]. Suspicious genetic changes have also been identified in chromosomes 14 and 3[7]. This is the description of a clinical case of a young patient with ARVD.

Case Report

A 25-year-old male, married patient from Santo André, São Paulo State, was admitted at Hospital do Coração at São Paulo in March, 2007, with a history of syncope during sexual intercourse. The patient was unable to inform on episode duration. After the syncope, the patient referred ventilation-dependent intense, oppressive precordial pain. The patient informed he was a triathlete, and not a smoker. The patient reported no family history of sudden death.

Physical examination was normal, except for frequent extra-systoles.

ECG at admission showed evidence of sinus rhythm, second degree bundle branch block (BBB), and frequent ventricular extrasystoles in LBBB pattern.

The patient was referred to the Coronary Care Unit, remained asymptomatic, and kept frequent, monomorphic extrasystoles. The following exams were performed:

- Transthoracic ECG: No findings
- Holter 24h: 1,443 monomorphic ventricular extrasystoles
- Ergometric test: Isolated, frequent monomorphic ventricular extrasystoles; supraventricular extrasystoles – isolated and rare on effort. Normal heart rate behavior and blood pressure.
- Cardiac MRI (Figure 1): Reduction in RV mid-anterior wall thickening with diskynetic movement, hyperintense signal in late phase after the injection of paramagnetic contrast medium - suggestive of ARVD.

The electrophysiologic study showed ventricles to be stable even under isoproterenol-sensitization and stimulation protocols at RV border and exit pathway; lack of signs of Brugada syndrome.

ARVD diagnosis was reached based on clinical and radiological data.

On day 4 after hospital admittance the patient was submitted to successful extrasystole ablation of RV exit pathway. The focus of arrhythmia was located at RV mid-septal area, close to the bundle of His.

The patient progressed in sinus rhythm, with no extrasystoles, and was discharged on sotalol 160mg/day, with indication for very close follow-up, and full restriction of extenuating physical activity. After a 5-month follow-up, the patient is asymptomatic, in sinus rhythm, presenting no extrasystoles and discreet RV dilation on control ECG.

Discussion

ARVD is characterized by LBBB-pattern ventricular arrhythmias. Morphologically, it is characterized by RV myocardial infiltration with fibrofatty tissue[7]. The infiltration usually occurs in the diaphragmatic, apical, and infundibular region – known as the "dysplasia triangle" – and may develop to dilation or aneurysm[2].

ARVD patients may present symptoms at any age, more

Key words

Arrhythmogenic right ventricular dysplasia; arrhythmia; heredity.

Mailing Address: Rogério Ferreira da Silva•
Rua Desembargador Eliseu Guilherme, 31/45 – Paraíso - 04004-030, São Paulo, SP - Brazil
E-mail: rfsilva@hcor.com.br
Manuscript received August 13, 2007; reviewed manuscript received September 07, 2007; accepted September 26, 2007.

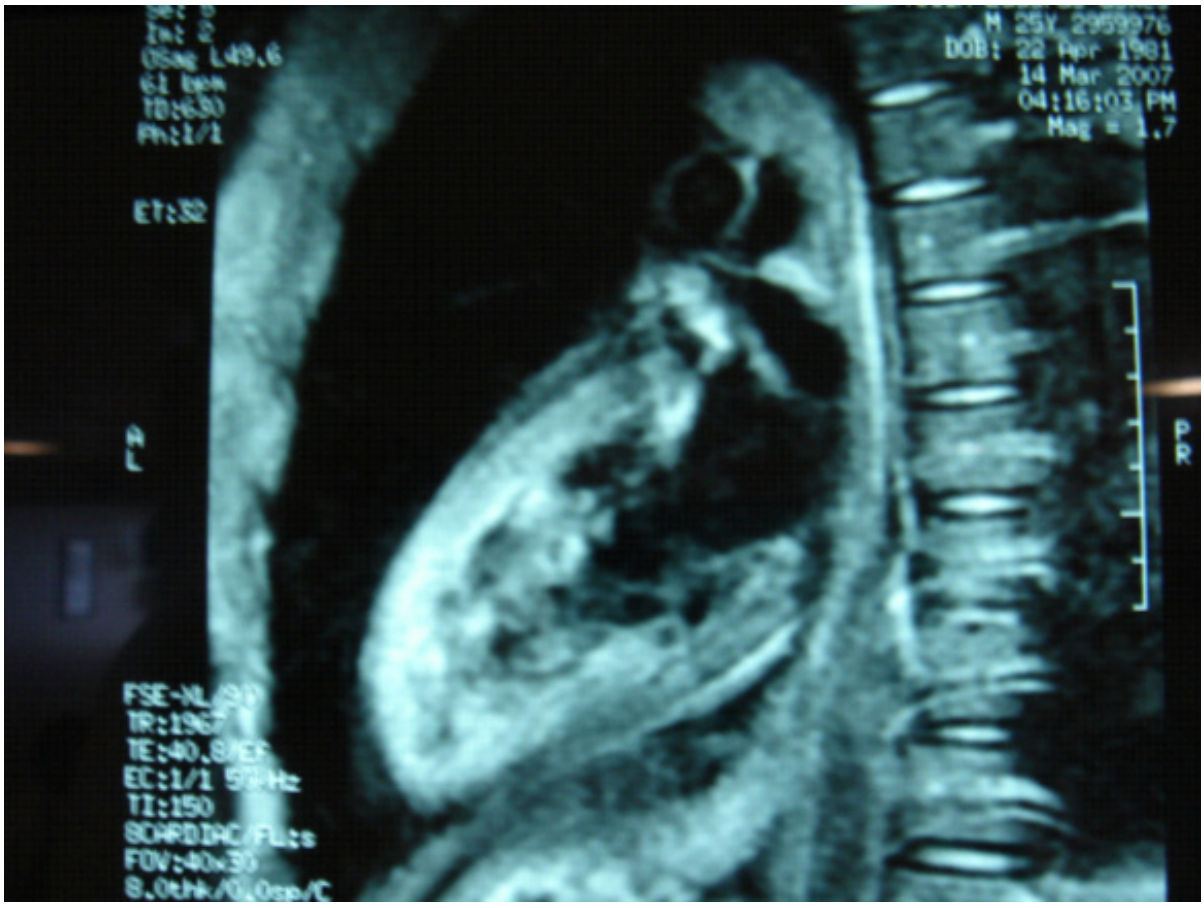

Figure 1 - Cardiac MRI.

commonly at young age, with predominance for male gender (ratio 3:1)[2].

Palpitations, fatigue and syncope are the most common symptoms. In some cases, cardiac arrest after physical effort may be the first presentation[2].

Estimated yearly mortality rate is 3% if not treated, and 1% under pharmacological treatment - implantable cardioverter defibrillator (ICD) excluded. Sudden death mechanism is the degeneration of ventricular tachycardia to ventricular fibrillation[3].

Fibrofatty islands generate macro-reentries, thus forming the arrhythmogenic substrate. Arrhythmia is induced by adrenergic stimulation as catecholamine infusion or physical activity[3].

ARVD may cause right ventricle or biventricular heart failure due to dilation and loss of contractile function[3].

Final diagnosis requires the histological finding of fibrofatty tissue[3]. In 1994, the European Society of Cardiology proposed a system of endpoints to help reaching diagnosis (Table I). Patients should present two major endpoints, one major and two minor, or four minor[3,8].

EGC typical finding is epsilon waves – "postexcitation" electrical potentials of small amplitude that occur at the end of the QRS complex. Epsilon waves are found in 30% of cases[3].

On ECG the most suggestive findings are RV dilation, with localized aneurysm and diskynesia[3].

MRI allows a morphofunctional analysis of ventricles and identifies adipose tissue. Some authors have suggested that MRI should be used to replace biopsy[9]. The endpoints that may be identified by MRI are: fibrofatty infiltration in RV with hyperintense signals, fibrofatty infiltration causing diffuse reduction of myocardial thickening, aneurysm or dilation of RV and of RV exit pathway, segmental contraction and global systolic or diastolic dysfunction[7].

Pharmacological treatment is the first choice option, through the administration of antiarrhythmics such as sotalol, verapamil, amiodaron, betablockers, propafenone - with variable therapeutic responses[2,7]. Extenuating physical activity should be avoided[2].

Radiofrequency ablation may be used for persistent or recalcitrant ventricular tachycardia, as well as for frequent tachyarrhythmias that trigger the implantable cardioverter-defribillator (ICD). The aim for ablation is to eliminate conduction pathways that perpetuate arrhythmia. Success has been reported from 30% to 65% of cases. New arrythmogenic foci may arise[2].

The proper time for ICD implant is debatable for patients on

Table 1 - Criteria for the diagnosis

Family history	
Major	- family history condition confirmed by necropsy or surgery
Minor	- family history or sudden death before 35 years of age, with suspicion of ARVD
ECG presenting depolarization or changes in repolarization	
Major	- epsilon waves or increase of QRS>100 ms in right precordial leads (V1-V3)
Minor	- late potentials in high resolution ECG - T wave inversion on right precordials of subjects under 12 years of age in the absence of right bundle branch block
Arrhythmias	
Minor	- ventricular tachycardia, sustained or not, in left branch bundle block fashion - frequent ventricular extrasystole over 1000/24 h (Holter)
Global or regional dysfunction and structural changes	
Major	- increased dilation and reduction in right ventricle ejection fraction, left ventricle little affected if at all - right ventricle aneurysm (akinetic or dyskinetic areas). Severe right ventricle dilation
Minor	- right ventricle mild global dilation or right ventricle ejection fraction reduction with left ventricle normal ejection fraction - right ventricle moderate segmental dilation - right ventricle regional hypokinesia
Tissue Characteristics of Wall	
Major	- presence of myocardial fibrous / fatty tissue evidenced by endomyocardial biopsy

BCRE - bloqueio completo do ramo esquerdo, VD - ventrículo direito; VE - ventrículo esquerdo.

optimized arrhythmics. When faced by recalcitrant arrhythmia in very young patients, the history of cardiac arrest and LV involvement should be considered[2].

ARVD is a progressive condition that follows an uncertain course. It may be the cause of the sudden death in young athletes, or an incidental finding in the necropsy of older patients[2]. Dalal et al[10] have described a series of 100 patients with ARVD and an average follow-up time of 6 years. From those, 66 survived (5 developed heart failure, 2 were transplanted, 44 implanted ICDs) and 34 did not survive (21 had sudden death as first presentation, and 10 during follow-up)[10].

In the case being reported, the patient presented LBBB-pattern, non-sustained tachycardia, frequent ventricular extrasystoles (higher than 1000/Holter 24h), RV diskynesia. The patient had 2 major endpoints added, and 1 major endpoint following the guidelines by the European Society of Cardiology. As for MRI endpoints, the following were observed: fatty infiltration of RV with hiperintense signals, reduction of myocardial thickening, and RV segmental change. Classic endpoints associated to MRI make ARVD diagnosis quite probable. The authors have concluded that MRI contributes for the diagnosis of ARVD.

Potential Conflict of Interest

No potential conflict of interest relevant to this article was reported.

Sources of Funding

There were no external funding sources for this study.

Study Association

This study is not associated with any graduation program.

References

1. Fontaine GH, Guiraudon G, Frank R. Stimulation studies and epicardial mapping in ventricular tachycardia: study of mechanisms and selection for sugery. In: Kulbertus HE, ed. Re-entrant arrhythmias: mechanisms and treatment. Baltimore: University Park Press, 1977. p. 334-50.
2. Anderson EL. Arrhythmogenic right ventricular dysplasia. Am Fam Physician. 2006; 73 (8): 1391-8.
3. Gemayel C, Pelliccia A, Thompson PD. Arrhythmogenic right ventricular cardiomyopathy. J Am Coll Cardiol. 2001; 38 (7): 1773-81.
4. Czarnowska E, Wlodarska EK, Zaleska T. Arrhythmogenic right ventricular cardiomyopathy (dysplasia): etiology, clinical presentation, diagnosis and treatment. Kardiol Pol. 2003; 58 (1): 58-63.
5. Sen-Chowdhry S, Lowe MD, Sporton SC, McKenna WJ. Arrhythmogenic right ventricular cardiomyopathy: clinical presentation, diagnosis and management. Am J Med. 2004; 117 (9): 685-95.
6. Marcus FI, Fontaine GH, Guiraudon G, Frank R, Laurenceau JL, Malergue C, et al. Right ventricular dysplasia: a report of 24 adult cases. Circulation. 1982; 65: 384-98.
7. Kayser H, van der Wall E, Plein S, Sivananthan MU, Bloomer TN, de Ross A. Diagnosis of arrhythmogenic right ventricular dysplasia: a review. Radiographics. 2002; 22: 639-48.
8. Corrado D, Buja G, Basso C, Thiene G. Clinical diagnosis and management strategies in arrhythmogenic right ventricular cardiomyopathy. J Electrocardiol. 2000; 33: 49-55.
9. Elias J, Tonet J, Fontaine G, Frank R. Displasia arritmogência do VD. Arq Bras Cardiol. 1998; 70 (6): 449-56.
10. Dalal D, Nasir K, Bomma C, Prakasa K, Tandri H, Piccini J, et al. Arrhythmogenic right ventricular dysplasia: a United States Experience. Circulation. 2005; 112: 3823-32.