The International Journal of Cardiovascular Imaging (2005)
DOI 10.1007/s10554-005-9018-y

Case Report

Evaluation and course of an unusual case of arrhythmogenic right ventricular dysplasia

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Received 30 June 2005; accepted in revised form 16 July 2005

Key words: arrhythmogenic right ventricular dysplasia, arrhythmogenic right ventricular cardiomyopathy, magnetic resonance imaging, right ventricle

Abstract

We report a case of a 42-year-old Caucasian man who presented with isolated right ventricular failure and atrial fibrillation without ventricular arrhythmia. In this report, we describe accurate evaluation by MR imaging confirmed by histopathologic findings as well as imaging progression of this unusual case of arrhythmogenic right ventricular dysplasia.

Introduction

Arrhythmogenic right ventricular dysplasia (ARVD) is a cardiomyopathy that is histopathologically characterized by fatty and fibrofatty replacement of the right ventricular myocardium [1]. Clinical features of arrhythmogenic right ventricular dysplasia are progressive right heart failure and ventricular arrhythmias of right ventricular origin.

Diagnosis of ARVD at early stages remains a clinical challenge [2]. In the diagnostic follow-up MR imaging may be used to characterize the morphologic and functional changes in this disorder. Fat infiltration of the right ventricle may be identified by black blood MR imaging [3, 4] but the reliability of this finding has not been verified [5, 6]. Right ventricular enlargement or focal aneurysms may be identified on MR cine images. In addition fibrosis of the myocardium may be identified on delayed enhancement MR images [7].

In this report, we describe the diagnostic work-up as well as MR imaging findings in a patient presenting with an atypical form of ARVD, consisting of progressive right sided failure with late onset of non-sustained ventricular tachycardia.

Case report

A 42-year-old Caucasian male patient with no prior history of cardiac symptoms presented with shortness of breath, nocturnal dyspnea, pedal edema and a deterioration of his functional class over a period of 2 years. His medical history was unremarkable, except for a borderline high blood pressure.

Physical examination was notable for a blood pressure of 128/92 mm of Hg. His heart rate was irregular at 40–50 beats per minute (bpm). The jugular venous pressure was approximately 20 cm of H_2O with absent '*a*' waves. Carotids were normal without bruits. Cardiac examination revealed a prominent right ventricular heave. Cardiac auscultation revealed a normal S1 and a widely split S2 which varied with respiration without any significant murmurs. The lung fields were clear to auscultation. The abdomen was soft and nontender with pulsatile hepatomegaly. The extremities were notable for a for 1+ pitting edema with normal peripheral pulses.

Chest X-ray showed enlargement of cardiac silhouette with clear lung fields. ECG revealed atrial fibrillation with a ventricular rate of 40 bpm, normal axis and a wide QRS consistent with right bundle branch block. Echocardiography demonstrated normal left ventricular function, massive enlargement of the right atrium, moderate right ventricular enlargement and dysfunction, mild to moderate tricuspid regurgitation. The right ventricular systolic pressure was within normal limits.

A VQ scan showed low probability of pulmonary thrombo-embolism and a chest CT scan showed no evidence of parenchymal or interstitial lung disease. Cardiac MR images were obtained on a 1.5 T scanner (CV/i, General Electric Medical Systems, Waukesha, WI). On ECG-gated cine MR images (steady state free precession SSFP pulse sequence, FIESTA©, General Electric Medical Systems, Waukesha, WI) there was diffuse global hypokinesis of the right heart. There was no evidence of regional contraction abnormalities. The right ventricular ejection fraction was 25%. The right atrial diameter was 9.3 cm and right ventricle diameter was 6.5 cm. There was also enlargement of the right ventricular outflow tract and moderate tricuspid regurgitation. The left heart chambers were normal in size with left atrial diameter of 3.6 cm and left ventricular diameter of 4.3 cm with ejection fraction of 55%. There was paradoxical motion of the interventricular septum consistent with increased right heart pressures or right bundle branch block. On T1-weighted images (fast spin echo sequence, TR/TE 845/5.5) there was no evidence of abnormal increased signal intensity that would otherwise correspond to fat in the anterior wall of the right ventricle (Figure 1a). Myocardial delayed enhancement images (inversion recovery prepared fast gradient echo blood suppression sequence, TR/TE 5.3/1.3) were obtained approximately 15 min following the administration of 0.2 mmol/kg intravenous gadodiamide contrast (General Electric Health Systems, Princeton, NJ). Delayed myocardial images indicated enhancement of the anterior and inferior right ventricular wall compatible with fibrosis or inflammation (Figure 1b).

Invasive testing was undertaken to confirm the results of non-invasive testing. Cardiac catheterization demonstrated no left to right shunt, normal left sided filling pressures with elevated right heart filling pressures. Right ventriculography demonstrated globally hypokinetic dilated right ventricle with no focal aneurysms or regional dysfunction. Endomyocardial biopsy taken from the myocardial border of the interventricular septum revealed mild to moderate fibrosis with only few focal areas of adipose tissue infiltration (Figure 1c) consistent with the diagnosis of ARVD.

Electrophysiologic testing revealed no inducible ventricular arrhythmia on programmed stimulation even with Isoproterenol. The patient was treated conservatively with diuretics and digoxin for right sided heart failure and ICD therapy was deferred due to the lack of ventricular arrhythmia.

One year later, a 24-h Holter recording revealed bardycardia with a mean heart rate of 53 bpm varying from 37 to 151 bpm. There were occasional multiform isolated ventricular complexes and runs of ventricular tachycardia. The longest and fastest run was 9 beats at a heart rate of 134 bpm (Figure 1d).

Repeated cardiac MR imaging revealed progression of enlargement of the right atrium to 9.5 cm and enlargement of the right ventricle to 7.3 cm. The left heart chambers were unremarkable. In view of the progression of right ventricular disease and the non-sustained ventricular tachycardia an implantable defibrillator was placed to prevent sudden death [8].

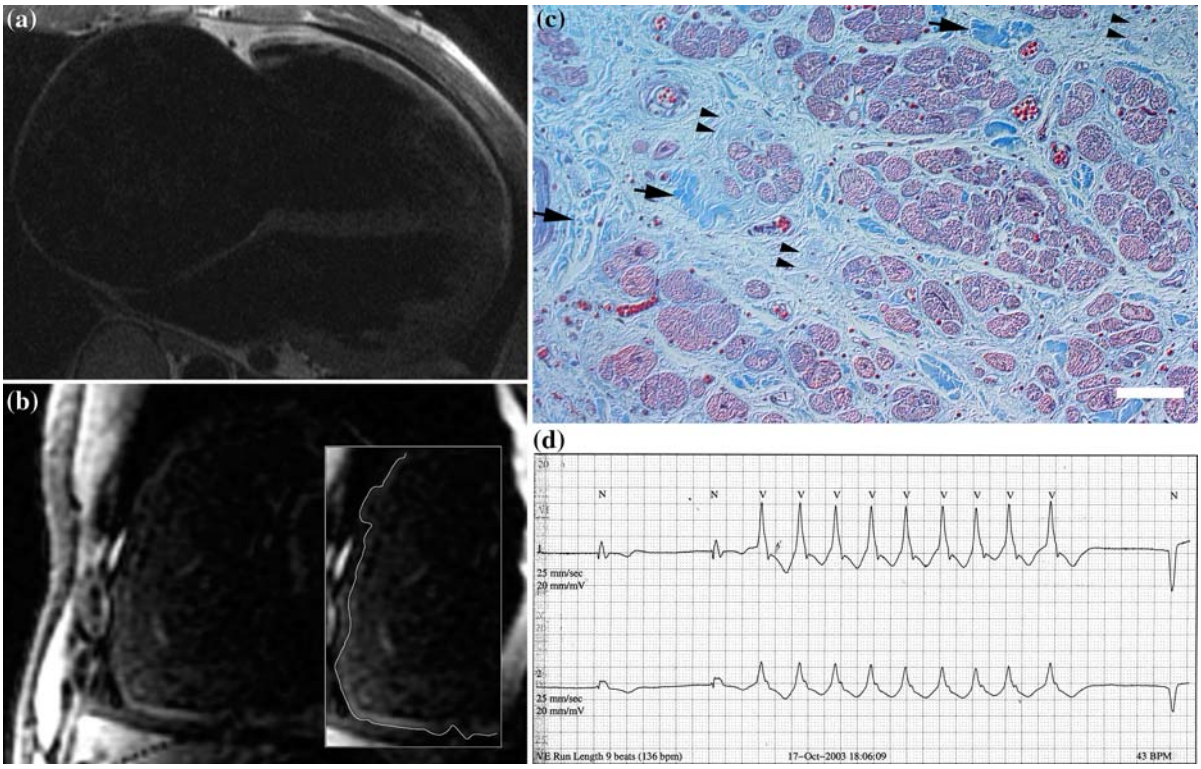

Figure 1. 42-year-old Caucasian man with atypical clinical presentation of ARVD: (a) T1-weighted MR image (fast spin echo sequence, TR/TE 845.1/5.5) showing no evidence of increased signal intensity (fat) in the anterior right ventricular wall. (b) Myocardial delayed enhancement images (inversion recovery prepared fast gradient echo blood suppression sequence, TR/TE 5.3/1.3) revealed enhancement of the anterior and inferior right ventricular wall consistent with myocardial fibrosis. The small image outlines the enhancement. (c) Light micrograph stained for collagen (Masson trichrome, Calibration bar = 100 μm) showing hypertrophied myocardium with abundant loose (arrowheads) and dense (arrows) collagen fiber bundles. These bundles are visible in the interstitial space, separating the myocytes. Note the absence of adipose tissue cells. (d) 24 h Holter monitor indicating a run of ventricular tachcardia of 9 beats.

Discussion

Diagnosis of arrhythmogenic right ventricular dysplasia is made according to major and minor criteria proposed by the Task Force of the Working Group on Cardiomyopathies [9]. Criteria most commonly observed in ARVD are ventricular arrhythmia, ECG repolarization changes and global or regional right ventricular dysfunction as well as morphologic abnormalities of the right ventricular free wall [10]. However, patients may be asymptomatic or symptoms may be limited to infrequent palpitations and occasional syncope.

MR imaging has evolved to a promising tool for evaluation of ARVD [11, 12]. In addition to highly accurate functional information about the right ventricle it provides unique information about myocardial characteristics, especially in differentiation of fatty and muscular tissue [13] that has been reported to be strongly associated with ARVD [3]. On T1-weighted MR-images fat appears with high signal intensity, whereas myocardium appears with intermediate signal intensity. However, these MR imaging findings were not considered as part of the Task Force criteria for ARVD [9] because of limited experience with MR imaging [5].

Following the initial evaluation, in our patient two major criteria of the Task Force criteria for ARVD were met: severe right ventricular dilatation in combination with reduced ejection fraction as well as histopathologic proven fibrofatty infiltration of the right ventricular myocardium. However, our patient is unique in that common clinical manifestations seen in patients presenting with ARVD were initially not present [2]. There was no spontaneous or inducible ventricular tachycardia, absence of fat signal on T1-weighted MR images, as well as no evidence of right ventricular regional contraction abnormality.

The histopathologic finding of only few fatty infiltrates in the right ventricular myocardium were consistent with the absence of fat signal on T1-weighted MR images. In addition, findings on delayed enhancement MR images correlated with the diffuse mild to moderate fibrosis of the right ventricle found in histopathology. Although delayed enhancement may be seen in other conditions of the right ventricle such as in inflammation [7], it is a promising tool for the non-invasive visualization of myocardial fibrosis. Recent reports suggest not only a roll in the diagnostic work-up but also in risk stratification due to the association of delayed enhancement and inducibility during electrophysiologic testing [14, 15].

In this case, MR imaging findings in delayed myocardial enhancement and T1-weighted MR images were consistent with the diagnosis of fibrofatty variant of ARVD confirmed by pathologic examination. Although there were no tachyarrhythmias on initial presentation a multidisciplinary diagnostic approach including cardiac MR imaging, electrophysiology and histopathology revealed the diagnosis of ARVD.

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