The Heart Surgery Forum #2009-1107
13 (1), 2010 [Epub February 2010]
doi: 10.1532/HSF98.20091107

Online address: http://cardenjennings.metapress.com

# Case Report: Nonarrhythmogenic Right Ventricular Dysplasia Presenting with Severe Right Ventricular Failure in an Adolescent

**Mustafa Pac**, Aysenur Pac, Tugcin Bora Polat, Sevket Balli, Nesrin Turhan, Dursun Aras, Mehmet Burhan Oflaz, Seref Kücüker

The Clinic of Pediatric Cardiology and Cardiac Surgery, Turkiye Yuksek Ihtisas Education and Research Hospital, Ankara, Turkey

## ABSTRACT

Right ventricular dysplasia is usually discovered by the presence of ventricular arrhythmia. As arrhythmia is an epiphenomenon, the first presentation of some cases can be primarily heart failure. We describe an adolescent girl who presented with progressive right heart failure and whose hallmark was fibrofatty replacement of ventricular muscle, especially of the right side, without ventricular arrhythmia. The patient was successfully treated by orthotopic heart transplantation.

## INTRODUCTION

Arrhythmogenic right ventricular dysplasia (ARVD) is characterized by the progressive replacement of myocardium with fatty or fibrofatty tissue. A fatty pattern in these patients represents the early stage of the disease before the appearance of fibrotic tissue [Marcus 1982; Webb 1986; Richardson 1996]. A large majority of patients with ARVD have histologic evidence suggestive of myocarditis, which produces fibrosis by involving cardiomyocytes and transforms the purely fatty tissue to the fibrofatty form. During this period, activation of neutrophils induced by the myocarditis may lead to ventricular arrhythmias and sudden death [Fontaliran 1991; Hoffman 1997].

It is possible that multiple attacks of myocarditis could lead to the destruction of an increasing number of myocardial cells involving both the right and the left ventricles, causing irreversible heart failure. For patients in whom ARVD has progressed to severe right ventricular or biventricular systolic dysfunction, treatment consists of current therapy for heart failure. In case of refractory heart failure, Glenn or Fontan procedures may be performed in selected cases, or the patients may become candidates for heart transplantation [Corrado 2000; Morita 2002; Gorgulu 2006; Gilljam 2009].

For the rare patients in whom a clear-cut pattern of ARVD is discovered by invasive or noninvasive tests of the right ventricle myocardium, patients do not have ventricular arrhythmias. In such patients, the arrhythmogenic substrate is present but silent [Girard 1997; Fontaine 1998; Blasco 2006].

We describe the case of an adolescent girl who presented with features of severe right ventricular failure and diagnosed fibrofatty-form nonarrhythmogenic right ventricular dysplasia that was managed with cardiac transplantation.

## CASE REPORT

A 16-year-old girl was admitted to the hospital with retrosternal chest pain, palpitation, and dizziness. A physical examination revealed a blood pressure of 110/90 mm Hg and a pulse rate of 110 beats/min. Her abdomen was distended with hepatomegaly and ascites, and she had bilateral leg edema. A telecardiogram revealed cardiomegaly, and an electrocardiogram (ECG) (Figure 1) showed atrial fibrillation with inverted T waves and an epsilon wave in leads V1 to V4.

Two-dimensional echocardiography showed regional right ventricular hypokinesia with dilation and thinning, as well as a large aneurysm at the apex of the right ventricle. Significant enlargement of right atrium with spontaneous contrast and thrombus formation at the posterior wall of the right atrium were also observed in the right atrium. The right ventricular shortening fraction and the ejection fraction were 7% and 15%, respectively. The results of the left ventricle echocardiographic evaluation remained normal except for a slightly impaired left ventricular contractility, which was measured as a shortening fraction and an ejection fraction of 28% and 58%, respectively.

Holter monitoring demonstrated 28 isolated monomorphic ventricular extrasystoles without any ventricular tachycardia, and atrial fibrillation was present during 24-hour recordings.

In accordance with these data, the patient received a diagnosis of ARVD, and a magnetic resonance imaging (MRI) evaluation was performed to confirm the diagnosis.

The MRI analysis revealed global right ventricular dilatation (becoming severe in the outflow tract), very decreased contractility with apical dyskinesis, subtricuspid pronounced telediastolic bulging, free anterior wall thinning, and multiple aneurysms; MRI showed no signs of fatty infiltration (Figure 2).

Symptoms attributable to right ventricular failure progressed despite intensive medical management during the subsequent 5 months. Therefore, we evaluated the patient for alternative surgical strategies to overcome cardiac failure.

*Received July 31, 2009; accepted September 4, 2009.*

*Correspondence: Professor Aysenur Pac, MD, The Clinic of Pediatric Cardiology, Turkiye Yuksek Ihtisas Education and Research Hospital, Kızılay S, No: 4, 06100, Sıhhiye, Ankara, Turkey; +90-312-3061724; fax: +90-312-3124120 (e-mail: aysepac@gmail.com).*

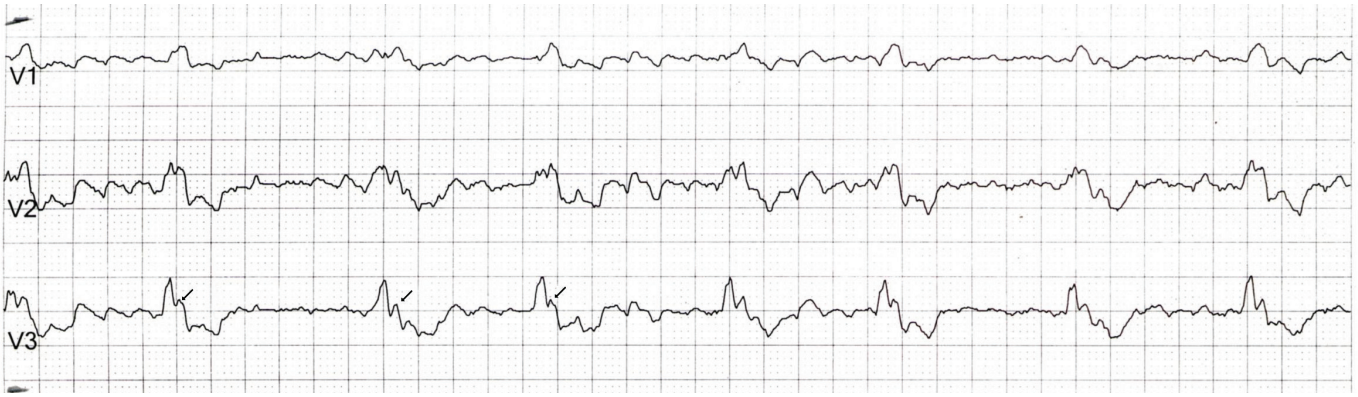


Figure 1. Electrocardiogram showing an epsilon wave (arrows) in lead of V1 to V3, with inverted T waves and atrial fibrillation.

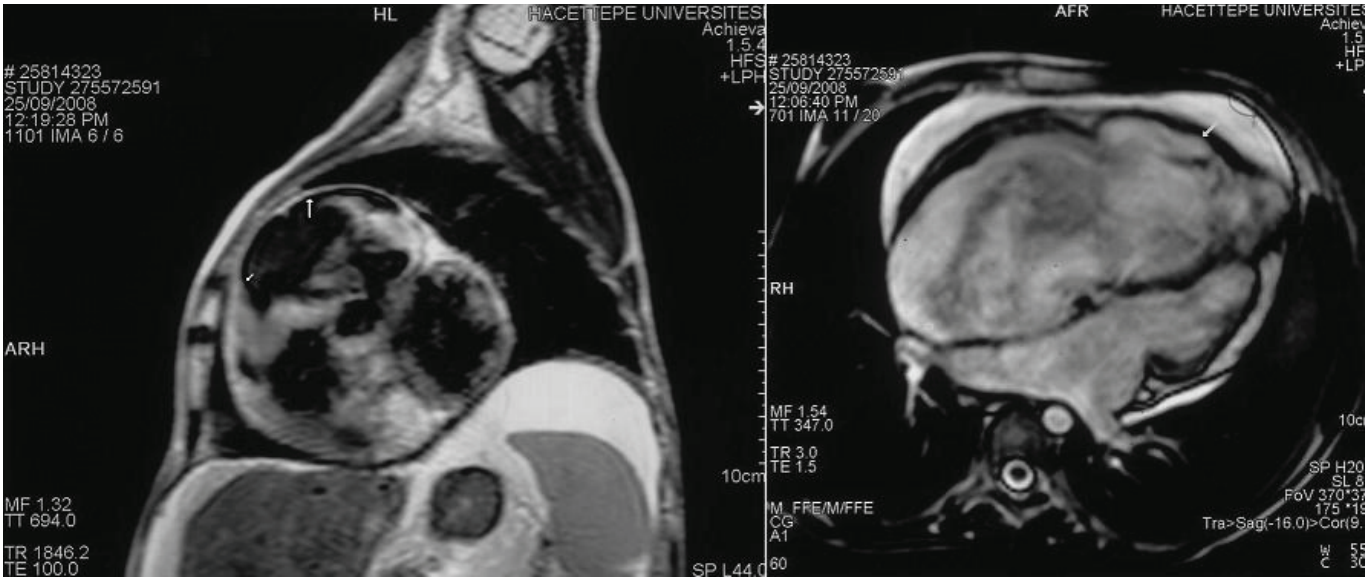


Figure 2. Apical dyskinesis (gray arrow) and right ventricle wall thinning and trabecular disarray (white arrows).

Cardiac catheterization was performed to determine which procedure would be appropriate for the patient. The mean pulmonary artery pressure was 22 mm Hg, and the pulmonary vascular resistance was 3.4 Wood units · $m^2$. Right ventricular cineangiography showed an aneurysmatic infundibulum with a poorly contracting right ventricle.

We agreed that heart transplantation would be the most appropriate treatment for this patient. One month later, she underwent an uneventful orthotopic transplantation.

A macroscopic examination revealed a dilated chamber with a thin-walled right ventricle, 1 to 3 mm in thickness. A transverse section revealed that the posterior free wall of the right ventricle had been entirely replaced with fibrous tissue. The left ventricle showed well-defined epicardial fat on the anterior and posterior walls, each of which measured 8 to 10 mm. Changes in the left ventricle predominated in the epicardial region (Figure 3).

A cardiac histologic evaluation revealed the typical pattern of ARVD, with extensive fibrofatty replacement of right ventricular myocardium, mainly in subepicardial and mediomural layers, with the replacement being regionally transmural with aneurysm formation. Surviving myocytes surrounded by fibrosis were embedded within the fatty tissue. Fibrofatty replacement of the left ventricular myocardium was also observed, mainly in the subepicardial layer (Figure 3).

At the 6-month follow-up, the patient continued to do well with no complaints, and routine surveillance biopsies of the endomyocardium have remained normal.

## DISCUSSION

The fibrofatty form of ARVD has histologic evidence suggestive of fibrofatty replacement and primarily affects the right ventricle. With time, the fibrofatty replacement may also involve the left ventricle, causing irreversible heart failure similar to the pathologic findings of end-stage dilated cardiomyopathies [Pinamonti 1992; Burke 1998; Dalal 2005].

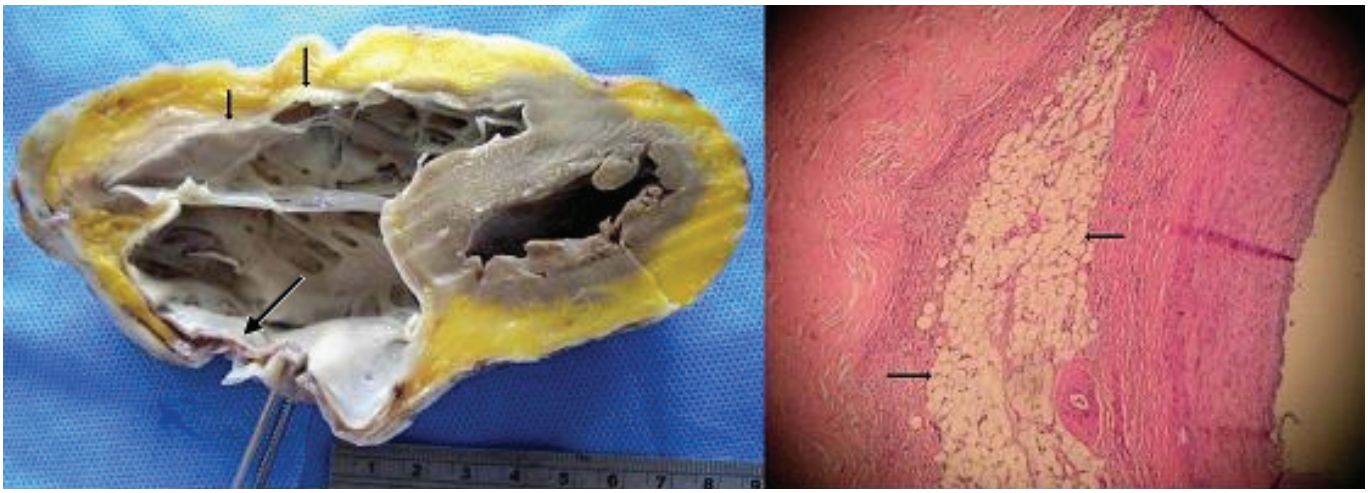

Figure 3. Macroscopic examination reveals the posterior free wall of the right ventricle to be thin and entirely replaced by fibrous tissue (arrows). A histologic evaluation shows evidence of the typical pattern of arrhythmogenic right ventricular dysplasia, with extensive fibrofatty replacement of the right ventricular myocardium (arrows), mainly in the subepicardial and mediomural layers (hematoxylin-phloxine-saffron staining; original magnification ×400).

ARVD rarely may present with progressive heart failure before arrhythmic complications become evident [Girard 1997; Fontaine 1998; Blasco 2006]; however, such patients may have ECG signs of ARVD and late potentials detected by signal averaging without having experienced arrhythmia or arrhythmia-related symptoms, as in our case.

MRI findings showed an excellent correlation with histopathologic results, suggesting a possible role in the evaluation and diagnosis of patients with suspected ARVD. However, cases presenting with severe and extensive signs indicating fatty replacement with right ventricular dysplasia, as was seen in our case, are very infrequent in the fibrofatty form of ARVD [Tandri 2003; Blasco 2006].

The specifics of the disease in the pediatric age group have not been well characterized because the mean age at diagnosis is 33 years [Dalal 2005]. Our patient had an unusual form of ARVD that had rapidly progressed to end-stage cardiomyopathy in the adolescent period.

We therefore performed cardiac catheterization to understand whether the patient would be a good candidate for a Glenn or Fontan operation, as had previously been performed in cases of refractory congestive heart failure with ARVD [Corrado 2000; Morita 2002; Gorgulu 2006; Gilljam 2009]. Because the mean pulmonary artery pressure and the pulmonary vascular resistance were slightly high, we considered the Glenn or Fontan procedure risky. Anyway, these procedures would not provide the required solutions to the following problems in this patient. First, although our patient did not experience arrhythmia or arrhythmia-related symptoms, ECG signs of ARVD were present that indicated an arrhythmogenic substrate, and these signs might have led to fatal arrhythmias during follow-up. Second, the patient had a slightly impaired left ventricular function; thus, we considered myocarditis to be progressing on the left ventricle.

Therefore, the patient underwent orthotopic heart transplantation. A cardiac histologic evaluation of the removed heart revealed fibrofatty replacement of the left ventricular myocardium, which retrospectively supported our decision regarding heart transplantation.

Besides our case, Cubero et al [2002] also reported this kind of situation. Similarly, their patient had a high pulmonary artery pressure and underwent cardiac transplantation because of refractory right heart failure. The patient died 1 week after the operation. Our patient underwent a successful cardiac transplantation and was discharged from the hospital with complete improvement in her condition.

We consider this case report important because it indicates that ARVD may cause severe right ventricular heart failure before arrhythmic complications become evident, the so-called nonarrhythmogenic right ventricular dysplasia. One should recognize, however, that such patients have a potential for left ventricular failure and fatal ventricular arrhythmias during follow-up. In this report, we have described, to our knowledge, the youngest patient to have undergone orthotopic heart transplantation for ARVD. We suggest that heart transplantation is the only way to change the natural evolution of and remit ARVD when cardiac failure occurs.

## REFERENCES

Blasco PB, Comas JG. 2006. Severe arrhythmogenic right ventricular dysplasia without fatty infiltration on magnetic resonance imaging: fibrofatty form. Pediatr Cardiol 27:476-7.

Burke AP, Farb A, Tashko G, et al. 1998. Arrhythmogenic right ventricular cardiomyopathy and fatty replacement of the right ventricular myocardium: Are they different diseases? Circulation 97:1571-80.

Corrado D, Basso C, Thiene G. 2000. Arrhythmogenic right ventricular cardiomyopathy: diagnosis, prognosis, and treatment. Heart 83:588-95.

Cubero JM, Gallego P, Pavon M. 2002. Arrhythmogenic right ventricular cardiomyopathy and atrial right-to-left shunt. Acta Cardiol 57:443-5.

Dalal D, Nasir K, Bomma C, et al. 2005. Arrhythmogenic right ventricular

dysplasia. A United States experience. Circulation 112:3823-32.

Fontaine G, Fontaliran F, Frank R. 1998. Arrhythmogenic right ventricular cardiomyopathies: clinical forms and main differential diagnoses. Circulation 97:1532-5.

Fontaliran F, Fontaine G, Fillette F, et al. 1991. Frontières nosologiques de la dysplasie arythmogene: variations quantitatives du tissu adipeux ventriculaire droit normal. Arch Mal Coeur 84:33-8.

Gilljam T, Bergh CH. 2009. Right ventricular cardiomyopathy: timing of heart transplantation in Uhl's anomaly and arrythmogenic right ventricular cardiomyopathy. Eur J Heart Fail 11:106-9.

Girard F, Fontaine G, Fontaliran F, et al. 1997. Catastrophic global heart failure in a patient with non-arrhythmogenic right ventricular dysplasia. Heart Vessels 12:152-4.

Gorgulu S, Nurkalem Z, Celebi A, et al. 2006. Unusual presentation of a patient with arrhythmogenic right ventricular dysplasia treated with a Glenn shunt. Int J Cardiol 113:410-3.

Hoffman BF, Feinmark SJ, Guo SD. 1997. Electrophysiologic effects of interactions between activated canine neutrophils and cardiac myocytes. J Cardiovasc Electrophysiol 8:679-87.

Marcus FI, Fontaine GH, Guiraudon G, et al. 1982. Right ventricular dysplasia: a report of 24 adult cases. Circulation 65:384-98.

Morita K, Takeuchi M, Oe K, et al. 2002. Perioperative management of Fontan operation for two patients with arrhythmogenic right ventricular dysplasia. J Anesth 16:169-72.

Pinamonti B, Sinagra G, Salvi A, et al. 1992. Left ventricular involvement in right ventricular dysplasia. Am Heart J 123:711-24.

Richardson P, McKenna W, Bristow M, et al. 1996. Report of the 1995 World Health Organization/International Society and Federation of Cardiology Task Force on the Definition and Classification of Cardiomyopathies. Circulation 93:841-2.

Tandri H, Calkins H, Nasir K, et al. 2003. Magnetic resonance imaging findings in patients meeting task force criteria for arrhythmogenic right ventricular dysplasia. J Cardiovasc Electrophysiol 14:476-82.

Webb JG, Kerr CR, Huckell VF, et al. 1986. Right ventricular abnormalities in arrhythmogenic right ventricular dysplasia. Am J Cardiol 58:568-70.