

Right Ventricular Dysplasia in an Asymptomatic Young Man: An Uncommon Case with Biventricular Involvement and No Known Family History

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A 33-year-old man had cardiomegaly on a routine x-ray examination. He was asymptomatic with no history of infarction, syncope, or palpitations. There was no family history of congenital heart disease or sudden death. Two-dimensional transthoracic echocardiography demonstrated marked enlargement of the right atrium and ventricle with severely depressed right and left ventricular function that was consistent with right ventricular dysplasia. The patient was treated with an angiotensin-converting enzyme inhibitor and did well for 6 months, but then developed symptomatic left-sided congestive heart failure. Short-

term improvement was obtained with intravenous inotropic therapy, but he continued to have progressive symptoms of heart failure. Approximately 7 months after his initial presentation, the patient underwent orthotopic heart transplantation for intractable congestive heart failure. Pathologic examination of the explanted heart established the diagnosis of right ventricular dysplasia with left ventricular involvement. This is an uncommon presentation of right ventricular dysplasia with biventricular involvement and no known family history. (J Am Soc Echocardiogr 2001;14:317-20.)

CASE REPORT

A 33-year-old man of Chilean origin was referred for evaluation of cardiomegaly that was discovered on a plain film examination for abdominal pain. He was completely asymptomatic at the time of presentation and had no family history of congenital heart disease or sudden death. Physical examination revealed a young, thin man who appeared to be healthy and in no apparent distress. His blood pressure was 110/74 mm Hg with a pulse rate of 68 bpm. His neck veins were flat. The apical impulse was hypokinetic, enlarged, and displaced laterally in the fifth left intercostal space. The right ventricle could be palpated at the left lower sternal border. The first heart sound was soft with a normal second heart sound. No murmurs of either mitral or tricuspid insufficiency could be heard.

A soft S3 gallop was present at the apex and right sternal border. Deep inspiration caused the gallop at the right sternal border to grow louder and softened the gallop at the apex.

Chest radiography showed cardiomegaly with no evidence of congestion. A 12-lead electrocardiogram revealed normal sinus rhythm with low voltage and epsilon waves (Figure 1). A 2-dimensional echocardiogram showed a severely dilated right atrium and right ventricle with severely reduced left and right ventricular systolic function (Figure 2). Results of plasma serum titers for *Trypanosoma cruzi* were negative. The patient was started on a low dose of an angiotensin-converting enzyme inhibitor and did well for 6 months, when he returned because of decreased exercise tolerance and bilateral pedal edema. He was admitted to the hospital, and cardiac catheterization demonstrated normal coronary arteries with a mixed venous oxygen saturation of 48% and a cardiac index of 1.4 L/min/m². The patient was treated with intravenous milrinone and was subsequently listed for cardiac transplantation. Three weeks later he underwent orthotopic heart transplantation for biventricular failure and is now doing well. Pathologic examination of the explanted heart revealed segmental areas of fibrofatty replacement of the right ventricle (Figure 3) and the lateral wall of the left ventricle (Figure 4).

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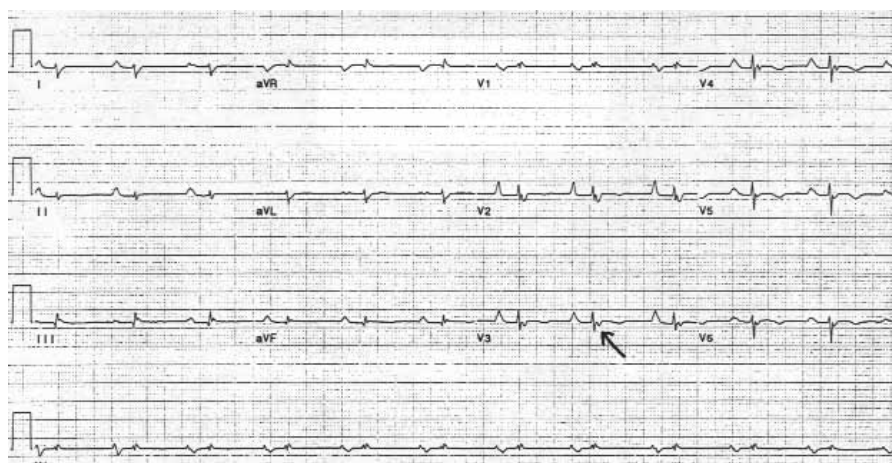


Figure 1 Standard 12-lead electrocardiogram reveals right atrial abnormality with first-degree atrioventricular delay and low QRS voltage. Note the T-wave inversions in the precordial leads and the epsilon-waves (*arrow*) following the QRS complexes that are most prominent in leads V₂ through V₄.

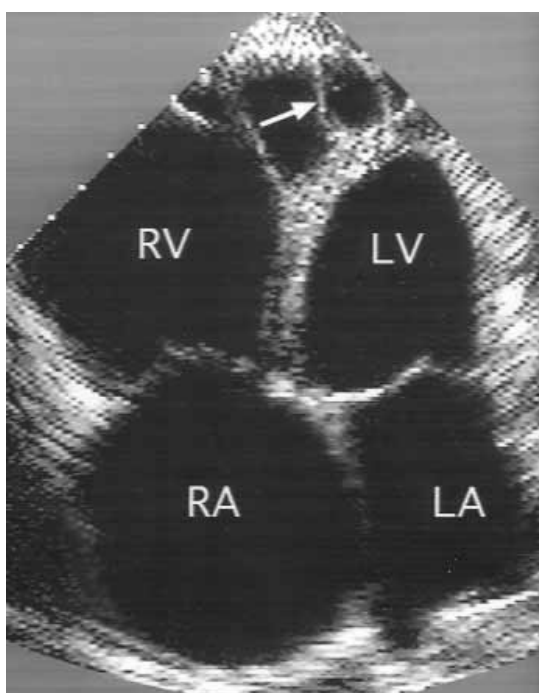


Figure 2 Transthoracic echocardiography in the apical 4-chamber view reveals a severely enlarged right atrium (RA) and right ventricle (RV). Note the heavy trabeculation and multiple septations with thinning of the right ventricular apex (*arrow*), which wraps around the left ventricular apex. LV, Left ventricle; LA, left atrium.

DISCUSSION

Right ventricular dysplasia is an idiopathic cardiomyopathy with a strong familial tendency¹ that has been mapped to several genetic loci.² Patients com-

monly present with ventricular arrhythmias (45%), congestive heart failure (25%), heart murmurs (10%), no symptoms (10%), or complete heart block (5%); 5% of patients have sudden death.³ Right ventricular dysplasia typically occurs in children or young adults and is rarely discovered in persons older than 40 years. The prevalence of right ventricular dysplasia is estimated to be 1 in 5000, and this may be an underestimate because many cases are unrecognized. Histopathology demonstrates fibrofatty infiltration of the right ventricle with less frequent involvement of the left ventricle.⁴

We report a case of right ventricular dysplasia in an asymptomatic young man with biventricular involvement and biventricular failure. The diagnosis was suggested by the echocardiographic appearance of the patient's heart with marked enlargement of the right ventricle and a dyskinetic right ventricular apex that wrapped around the left ventricular apex.⁵ Given that the patient had emigrated from a country with a high incidence of *T. cruzi* infection, we confirmed that he did not have Chagas' disease with negative results of plasma serum antibodies. The patient was in the proper age group for initial presentation of right ventricular dysplasia, and the fact that he had no symptoms at the time was not inconsistent with the diagnosis, as above.³

The explanted heart weighed 371.5 g. The right ventricle was dilated, and there were segmental areas in the right ventricle where the wall was thin and translucent. There was a focal pale area in the lateral wall of the left ventricle. Microscopically, the right ventricle showed areas that were replaced by fibrofatty tissue and vacuolated myocytes trapped in fi-

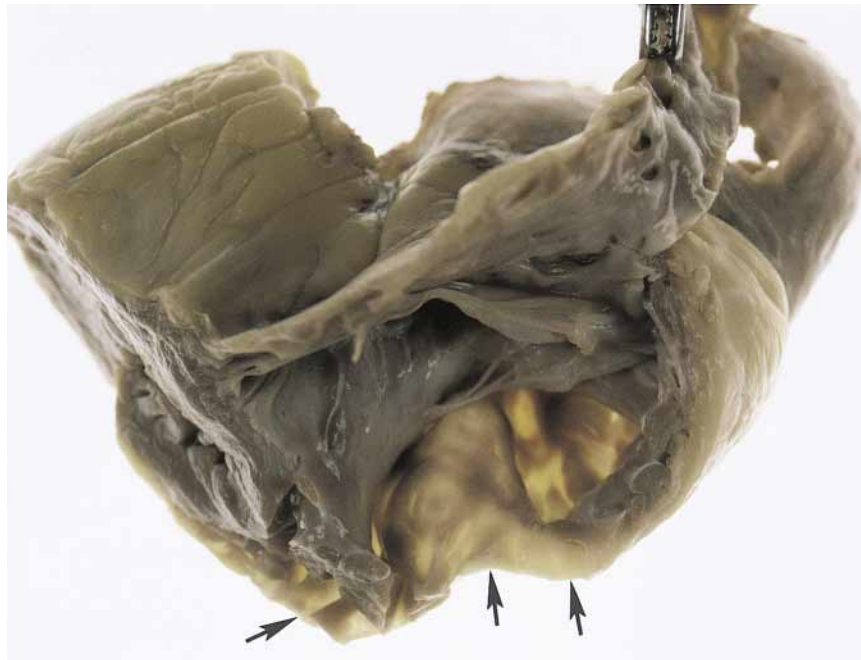


Figure 3 Transilluminated gross pathologic specimen of the patient's heart viewed from the right ventricular apical aspect. Note the extensive thinning of the right ventricle along the lateral and apical regions with multiple redundant outpouchings at the apex, as shown by the *arrows*.

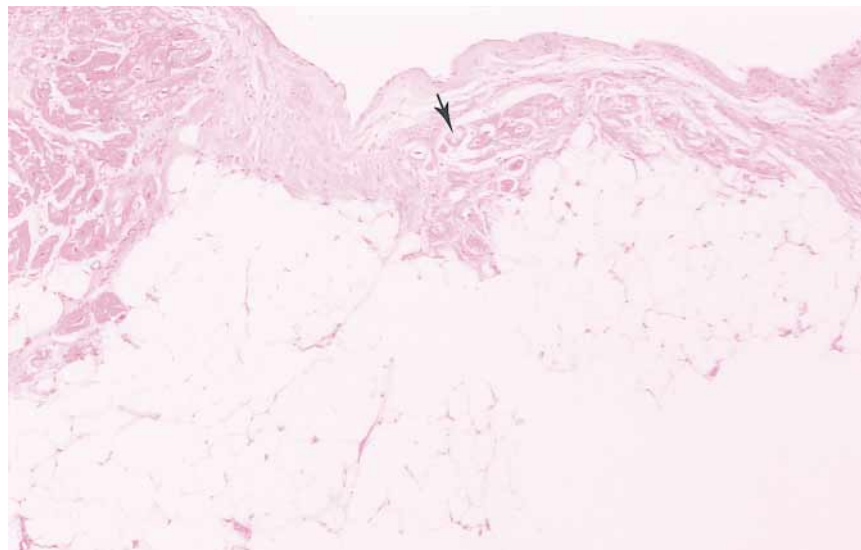


Figure 4 This photomicrograph (hematoxylin-eosin, $\times 120$) shows a section of the left ventricular free wall with the endocardium at the *top* of the figure and the epicardium at the *bottom*. Note that nearly all of the myocytes have been replaced by fibrous and fatty tissue. The fibrous tissue can be seen entrapping a few degenerating myocytes, as shown by the *arrow*.

brous tissue with no associated chronic inflammation. The left ventricle showed patchy areas of replacement fibrosis and a focal area in the lateral wall with fatty replacement. Basso et al⁶ found that biven-

tricular infiltration is most often associated with a fibrofatty pattern and that isolated right ventricular infiltration has a predominantly fatty pattern. Biventricular fibrofatty infiltration has been well-described⁴

and occurs in as many as 50% of cases of right ventricular dysplasia.³ However, this particular case is different because the patient was asymptomatic at the time of presentation, despite documented biventricular failure. In retrospect, his abdominal pain may have been caused by venous congestion from right ventricular failure, though the abdominal pain resolved and did not recur with treatment by H₂-receptor antagonists. In addition, the patient could not identify any first-degree family members with a history of congenital heart disease or sudden death. Even now, 2 years after his initial diagnosis, there has been no further detection of heart disease in his family, despite screening with echocardiograms of his siblings and electrocardiograms of all of his first cousins. To our knowledge, no antemortem diagnosis of a case of right ventricular dysplasia with biventricular involvement and with no family history has been reported.

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