

Isolated Form of Spongy Myocardium

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Noncompaction of the ventricular myocardium sometimes referred to as «spongy myocardium», is a rare congenital cardiomyopathy resulting from an arrest in normal endomyocardial embryogenesis. The characteristic echocardiographic findings of this disease consist of multiple myocardial trabeculations and deep intertrabecular recesses communicating with the left ventricular cavity. Familial occurrence has been observed. We present an illustrative case of isolated noncompaction of the ventricular myocardium in a 16-year-old patient, with the typical clinical and echocardiographic features of the disease. The literature on the topic is reviewed.

Key words: *Spongy cardiomyopathy. Noncompaction ventricular myocardium.*

Forma aislada de miocardiopatía esponjiforme

Una forma muy infrecuente de miocardiopatía congénita, resultado de la interrupción en el normal desarrollo embrionario endomiocárdico, consistente en la no compactación del miocardio ventricular, es la «miocardiopatía esponjiforme». Las características ecocardiográficas de esta enfermedad consisten en múltiples trabeculaciones y recesos que comunican directamente con la cavidad ventricular. Existe una alta incidencia familiar de esta enfermedad. Presentamos un caso ilustrativo de la forma aislada de esta enfermedad, no asociada a otras anomalías, en un paciente de 16 años de edad con las manifestaciones clínicas y ecocardiográficas típicas de este proceso patológico. Revisamos a continuación la bibliografía de esta afección.

Palabras clave: *Miocardiopatía esponjiforme. Miocardio no compactado.*

INTRODUCTION

Spongiform cardiomyopathy, also known as «non-compacted myocardium,» is an infrequent form of congenital cardiomyopathy resulting from a disturbance in endomyocardial embryogenesis.

The diagnosis of this disease is based on typical echocardiographic findings, such as intramyocardial recesses and trabeculations. Clinical symptoms include ventricular systolic dysfunction, fundamentally of the left heart, including heart failure, ventricular arrhythmias, and embolism. Its clinical aspects and prognosis are similar to those of idiopathic dilated cardiomyopathy. We report a case of isolated spongiform cardiomyopathy with no other associated anomalies.

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CLINICAL CASE

A 16-year-old male with heart failure refractory to conventional treatment was referred to our center for heart transplantation assessment.

His family history included the sudden death of his mother at the age of 32 years, although there was no causal diagnosis of her death. No cardiological or genetic study was made of the father or other first-degree relatives.

He was diagnosed as having non-obstructive hypertrophic cardiomyopathy with severe systolic dysfunction in the reference hospital one year earlier. The patient was referred to our center for heart transplantation assessment due to progressive heart failure that eventually became evident at rest (grade IV). His medical history revealed no evidence of arrhythmia or embolic episodes.

The clinical picture was heart failure affecting the left heart predominantly. His heart failure had progressed in recent months in spite of treatment with furosemide (80 mg/d), spironolactone (25 mg/d), digoxin (0.25 mg/d), enalapril (10 mg/d), and carvedilol (6.25 mg/12 h).

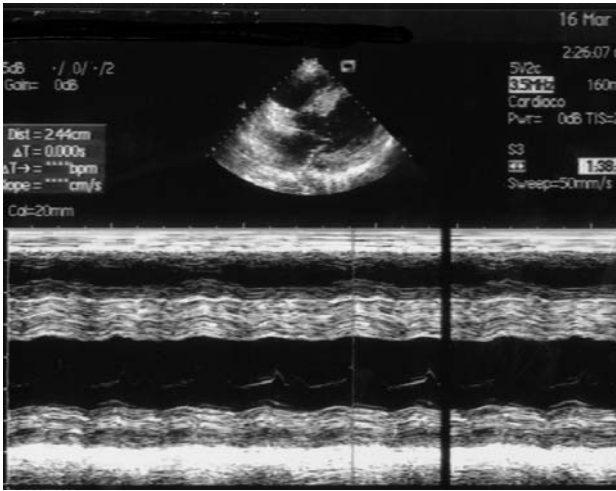


Fig. 1. Bidimensional echocardiographic image, through the long axis of the parasternal plane, showing myocardial thickening with severe systolic dysfunction of the left ventricle.



Fig. 2. Bidimensional echocardiographic image (four-chamber apical view) revealing recesses and trabeculae restricted mainly to the apical segment.



Fig. 3. Cross-section of the heart of the receptor in which the left ventricular recesses and trabeculae are visible.

The physical examination confirmed his poor general condition and systemic hypoperfusion. His blood pressure was 105/65 mm Hg. Auscultation disclosed rhythmic heart tones, 120 beats per minute, no murmurs, a third heart sound, and wet-sounding rales in both pulmonary bases.

Laboratory tests found low blood sodium (127) and mild kidney dysfunction (creatinine 1.8 mg/dl). The blood tests and coagulation study were normal. The electrocardiogram showed sinus tachycardia with left ventricular enlargement and isolated monomorphic extrasystoles; QRS duration was 110 ms.

Moderate cardiomegaly and signs of high venous and capillary pressure were evident in the chest X-ray exam.

Echocardiography disclosed dilation of the left ventricle, severe systolic dysfunction, a 12% ejection fraction, and 24-mm septum (Figure 1). In the four-chamber apical plane, the endocardium had recesses and prominent intramyocardial trabeculations in the apical segments (Figure 2).

The patient's clinical evolution was unfavorable and he required high-dose inotropic medication from the moment of admission. He underwent heart transplantation during this hospital stay. Although he had an episode of IIIA rejection, his later evolution was very satisfactory and he was asymptomatic and in very good clinical condition at 10 months of follow-up.

Pathological study of the heart confirmed the diagnosis of spongiform cardiomyopathy. The pathological evidence correlated closely with the recesses and trabeculations observed in the echocardiographic study (Figure 3).

DISCUSSION

Among the cardiomyopathies, a group of diseases of the myocardium that cause poor myocardial function, spongiform cardiomyopathy is very infrequent. It has been included in the group of unclassified cardiomyopathies.¹ In spite of its rarity, spongiform cardiomyopathy has been diagnosed with increasing frequency in the last years.² The disease, initially described in children,³ has also been observed in adult patients. This trend toward more frequent diagnosis may be due to the relatively ease with which the disease is recognized in bidimensional echocardiography, which reveals a characteristic image of myocardial recesses communicating directly with the ventricular cavity in the absence of other anomalies.⁴ These images coincide with histopathological findings⁵ (Figure 3).

Although the condition can affect the right ventricle and interventricular septum, in the few series published in the literature, the left ventricle is usually involved, and biventricular involvement occurs in 40% of cases.³

As mentioned above, the disease is characterized by systolic and diastolic left ventricular dysfunction, with clinical manifestations of heart failure.⁶

There are two variants of the disease:⁵ The least frequent form is isolated and not associated with any cardiac or extracardiac malformation. The other form is cardiomyopathy with facial malformations (prominent forehead, bilateral strabismus, micrognathia, and cleft palate) and frequent Wolff-Parkinson-White syndrome, particularly in children. Arrhythmias can occur, such as supraventricular arrhythmias mediated by pathways related with the Wolff-Parkinson-White syndrome, and ventricular arrhythmias whose electrophysiological mechanism has not yet been determined.⁵

The genetic basis of this disease⁷ is not fully known, but both the isolated variety and the variety associated with other malformations are related with a mutation of the *G 4,5* gene in Xq 28. The condition may be associated with Barth's syndrome (neutropenia, impaired growth, increased organic acids in urine, low concentrations of carnitine and mitochondrial anomalies), Emery-Dreifus muscular dystrophy, and myotubular cardiomyopathy. Familial occurrence is high, 44% of cases in the largest series.⁵ In the case of our patient, the cause of death of his mother was not determined and cardiological and genetic studies were not made of any other first-degree relative.

There is little information on the prognosis and management of these patients.⁸ In the largest series published to date, there is a parallel relation between the natural history, treatment, and dilated cardiomyopathy of idiopathic origin. The mortality 3 at 6 years is 80%. The disease is managed⁹ with conventional measures for heart failure, including anticoagulation and an-

tiarrhythmic medications. Automatic defibrillator implantation has been reported. Finally, heart transplantation is indicated, in accordance with current guidelines.

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