Case Report

Isolated Noncompaction of the Myocardium

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Noncompaction of the myocardium (NCM) is a rare congenital heart defect. It was first described 15 years ago, and few cases are published. The purpose of this study is to describe a NCM case. Clinical findings and imaging test results of a 37-year-old female patient with isolated NCM are presented. The patient complained of palpitations; her physical examination revealed extrasystoles, and her 12-lead electrocardiogram showed ventricular bigeminy. Three-dimensional Doppler echocardiography revealed numerous prominent trabeculations with deep intertrabecular recesses with blood flow which communicated with the ventricular cavity, which were more intense in the septal apical region. Cardiac magnetic resonance imaging corroborated the echocardiographic findings. The clinical presentation and the patient's laboratory test results confirmed the diagnosis of isolated NCM. The knowledge of the echocardiographic findings of this disease enables an early diagnosis and a more adequate treatment.

Introduction

During early embryonic development, the heart has a porous aspect because of a network of interwoven fibers that forms trabeculations separated by deep recesses which communicate with the ventricular cavity[1].

Between weeks 5 and 8 of embryonic life, this myocardium undergoes compaction proceeding from the epicardium to the endocardium and from the base to the apex, which will determine the normal aspect of the cardiac muscle. The coronary circulation develops during this process and the intertrabecular recesses are reduced to capillaries. This compaction occurs more significantly in the left ventricle than in the right ventricle[2,3].

Noncompaction of the myocardium (NCM) is a rare congenital heart defect initially described by means of autopsy in 1932. In the 1980's the first few studies using echocardiographic criteria for the diagnosis of this condition were published[4]. The arrest in the process of myocardial compaction results in the persistence of trabeculations and deep recesses which communicate with the ventricular cavity, a manifestation that is characteristic of this disease.

Key words

Heart defects, congenital; heart / embriology; ventricular dysfunction.

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The left ventricle is always involved, and biventricular involvement occurs in less than 50% of the cases, with apical predominance[1].

According to echocardiographic criteria, the prevalence is 0.014%. However, these figures may likely be underestimated, because the majority of the studies were conducted in tertiary care hospitals and in symptomatic patients.[2] Men are more often affected than women, and familial occurrence may be present in up to 44% of the cases[1,3,.5].

We describe the main clinical and laboratory test findings of a young female patient with isolated noncompaction of the myocardium according to echocardiographic criteria.

Case Report

Female, 37 years old, physician, white, single, catholic, born in the city of Barra Mansa (State of Rio de Janeiro) and residing in Sao Paulo, was admitted on October 27, 2005 complaining of intermittent palpitations for two weeks. She reported that in a previous cardiological assessment during one of these episodes of palpitations, supraventricular paroxysmal tachycardia (SVPT) that reverted with vagal maneuvers was identified. She denied smoking, alcohol consumption or relevant family history. Physical examination was normal, except for the presence of frequent extrasystoles.

She underwent a 12-lead resting electrocardiogram that showed ventricular bigeminy. Her chest radiographs (posteroanterior and left lateral views) were normal. Transthoracic three-dimensional echocardiogram demonstrated numerous prominent trabeculations with deep intertrabecular recesses and absence of other cardiac anomalies. Color Doppler showed blood flow into these recesses, communicating with the ventricular cavity. These trabeculations were more numerous and prominent in the septal apical region of the left ventricle (Fig. 1). Left ventricular (LV) diastolic dysfunction was also diagnosed. The patient underwent magnetic resonance imaging (MRI), whose findings corroborated those of the echocardiogram (Fig. 2).

According to clinical criteria and results of laboratory tests, isolated noncompaction of the myocardium was diagnosed. Amiodarone at usual doses was administered with maintenance of sinus rhythm and absence of ventricular extrasystoles.

As a complement to the cardiovascular risk stratification, the patient underwent an electrophysiological study (EPS) and an exercise test, which showed normal results.

On November 8, 2005 the patient was discharged in good clinical condition, asymptomatic and was advised to repeat the EPS one year later due to the risk of sudden death.

The patient was advised to ask all her first-degree relatives to undergo cardiological assessment and two-dimensional

Doppler echocardiography.

S.A.C. has been followed on an outpatient basis for three months and remains asymptomatic.

Discussion

NCM was first described in association with other congenital heart defects (anomalous origin of the left coronary from the pulmonary trunk, obstruction of the right and left ventricular outflow tract)[1].

Noncompaction is believed to be due to the fact that the ventricles are exposed to very high pressures during intrauterine life. Isolated NCM is even rarer and its cause remains not fully known[1,6,7].

Sporadic and familial forms of NCM have been described. In studies involving isolated NCM predominantly in children, familial occurrence was observed in half of the patients. Studies on isolated NCM in adults reveal an 18% familial recurrence. Although the genes responsible for the sporadic forms have not been identified, several genes related to the disease have been described in familial cases[2,5].

Patients may remain asymptomatic throughout their lives or, more commonly, present the following clinical manifestations: LV systolic dysfunction resulting from microcirculation dysfunction and low subendocardial perfusion due to an increased oxygen demand determined by the isometric contraction of the endocardium and myocardium that extend to the intertrabecular recesses; LV diastolic dysfunction caused by both an alteration in relaxation and diastolic restriction provoked by the numerous trabeculations; cardiac arrhythmias, such as atrial and ventricular fibrillation; atrioventricular or bundle branch blocks; Wolff-Parkinson-White syndrome; and embolic complications resulting from thrombus in the atria or intertrabecular recesses[1,2,7,8].

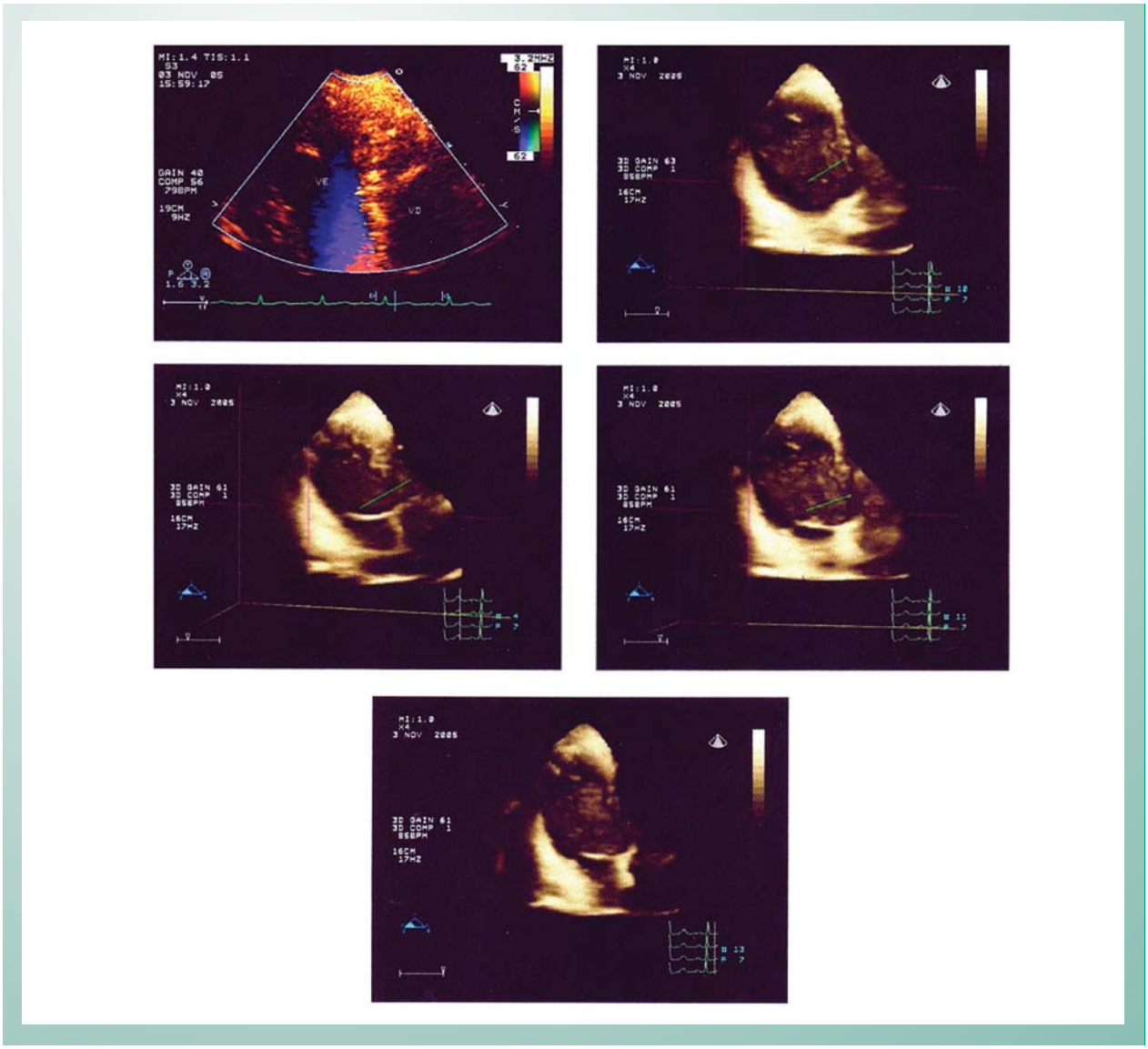

Fig. 1 - *Echocardiography showing myocardial trabeculations and recesses.*

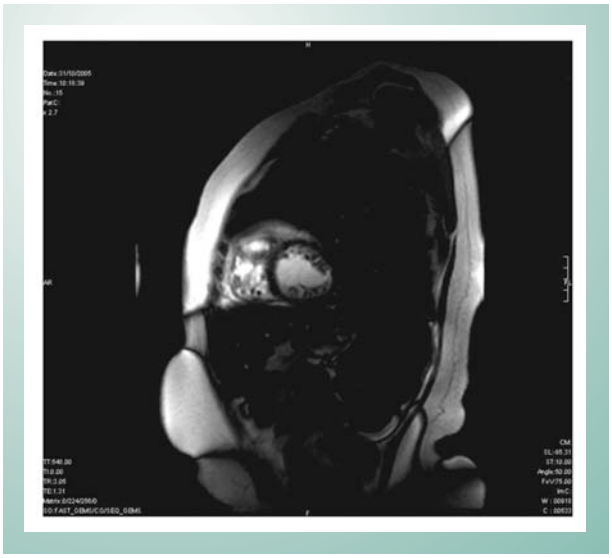

Fig. 2 - *Magnetic resonance imaging: increased myocardial trabeculations.*

Doppler echocardiogram is the most important laboratory test for the diagnosis of NCM. The criteria used for the diagnosis are: 1) absence of cardiac anomalies, as well as of anomalies in the aortic and pulmonary valves or in the coronary arteries; 2) numerous prominent trabeculations with deep intertrabecular recesses, and thickening ratio between the compacted and noncompacted layers > 2; 3) identification of blood flow directly from the ventricular cavity into the intertrabecular recesses; 4) involvement mainly of the mid-lateral, mid-lower or apical regions of the LV. Isolated NCM is diagnosed in the presence of three of these criteria[1,2,3,7,9]. The left ventricle is always involved and biventricular involvement occurs in less than 50% of the cases[1,2,10].

This patient presented palpitations, and a diagnosis was made of SVPT and ventricular bigeminy. Echocardiogram revealed diastolic dysfunction, which probably contributed to the occurrence of bigeminy and to the increased pressure in the left atrium which, in turn, may determine the onset of supraventricular arrhythmias. The echocardiographic findings were typical of NCM and since the four criteria previously described were present, the isolated form was characterized.

In two other case reports of NCM, MRI was used to confirm the diagnosis[11,12]. This imaging method may be used for the diagnosis and provides convincing evidences in cases in which the echocardiographic findings are inconclusive. Genetic tests for known mutations may provide additional data for counseling and research[13,14,15,16].

Differential diagnosis includes: prominent normal myocardial trabeculations (< 3); false tendons and aberrant bands; cardiac tumors and LV apical thrombus; apical hypertrophic cardiomyopathy; dilated cardiomyopathy; arrhythmogenic right ventricular dysplasia; and endocardial fibroelastosis[1,2,14,17].

Treatment depends on the clinical presentation. In cases of congestive heart failure, the treatment is similar to that of other causes. Some authors suggest electrophysiologic study and 24-hour Holter monitoring for patients in the initial assessment and annually, due to the risk of sudden death. Anticoagulation is indicated for all patients, regardless of the identification of intracardiac thrombus. Sports competitions are forbidden and pregnancy should be avoided[2,4,13,17].

The prognosis is widely variable, ranging from cases in which patients remain asymptomatic throughout their lives to cases with rapid deterioration of the cardiac function, causing early death[1,2,18].

First-degree relatives of patients with NCM should undergo Doppler echocardiography as a screening test[1,2,4]. Such a recommendation was made to the patient, because the early diagnosis enables a better risk stratification (including sudden death) and treatment.

We conclude that the echocardiographic findings enable an early diagnosis and better treatment of NCM.

Potential Conflict of Interest

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

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