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Case Report

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Left ventricular noncompaction apropos of a case

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Acronyms
LVN: Left ventricular noncompaction
ACD: automatic cardioverter-defibrillator
LV: left ventricle

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ABSTRACT

Left ventricular noncompaction, also known as "spongiform" cardiomyopathy, is a rare congenital anomaly of myocardial morphology, with deep intertrabecular recesses freely communicated with the ventricular cavity. The case of a 61 year old man, who was admitted to the Cardiology Service for syncope study, is presented. During his admission he underwent an echocardiogram and an isolated left ventricular noncompaction was diagnosed. It was decided to implant an automatic cardioverter-defibrillator in order to prevent potentially fatal malignant ventricular arrhythmias that might trigger his syncopes, after which the patient has progressed favorably.

Key words: Left ventricular noncompaction, spongiform cardiomyopathy, Arrhythmias, Sudden death

Miocardiopatía no compactada a propósito de un caso

RESUMEN

La miocardiopatía no compactada, también conocida como «espongiforme», es una anomalía congénita infrecuente de la morfología del miocardio, con recesos intertrabeculares profundos comunicados libremente con la cavidad ventricular. Se presenta el caso de un hombre de 61 años que fue ingresado en el Servicio de Cardiología para estudio por síncopes. Durante su ingreso se le realizó un ecocardiograma y se diagnosticó una miocardiopatía no compactada aislada. Se decidió implantar un cardiodesfibrilador automático con el objetivo de prevenir arritmias ventriculares malignas, potencialmente mortales, como causa de sus síncopes, tras lo cual el paciente ha evolucionado favorablemente.

Palabras clave: Miocardiopatía no compactada, Miocardiopatía espongiforme, Arritmias, Muerte súbita

INTRODUCTION

Left ventricular noncompaction (LVN), or spongiform cardiomyopathy, isolated, is a rare disease. It is characterized by alterations of the myocardial wall, with prominent trabeculae and deep intertrabecular recesses,

which give as a result a coarse myocardium with two layers. It can take place in the context of other genetic diseases with relative frequency; however, when presented in isolation is considered a rarity[1].

It was first described in the mid-eighties[2], but it was not until 2006 that the American Heart Association classified it as a disease[1].

It is between 1.4 and 2.7 cases per 1000 echocardiographic studies[3-5]. Because of its high association with sudden death, its diagnosis should be always taken into account by doctors, in order to implement therapeutic measures to avoid this complication[5].

In this article is presented a patient treated at the Department of Cardiology of the *Hospital Hermanos Ameijeiras*, Havana, Cuba; and the main clinical, diagnostic, therapeutic and prognostic features of this disease are summarized.

CASE REPORT

A 61-year-old man, black skin and hypertension-controlled for two years, was admitted for presenting episodes of loss of consciousness. He exposed that these began 13 years ago and they had happened to about ten, spaced over time, without any studies to determine their cause. The last two were six days prior to admission.

His loss of consciousness had been sudden, without aura, or palpitations; although in some occasions he presented previous dizziness. They were not accompanied by involuntary movements, and they lasted about a minute, leaving no sequels. No triggering factor was identified. These episodes occurred equally at rest or associated with physical exertion. The patient denied other symptoms during the questioning.

The physical examination revealed a systolic ejection murmur, more intense in the second and third left intercostal space, of intensity 2/6, without thrill or neck or armpit irradiation. Its intensity increased with the Valsalva maneuver and it decreased with the squatting position. Heart sounds were rhythmic, with no third or fourth noises, heart rate of 72 beats per minute, blood pressure of 118/75 mmHg and 18 breaths per minute. Arterial pulses were present and had good intensity.

The complete hemogram and coagulogram, the ionogram, and all the blood tests in general were normal; the chest X-ray showed a normal cardiothoracic index and no signs of pulmonary congestion, and the electrocardiogram showed the existence of a left bundle branch block.

The echocardiogram allowed to identify an increase in the thickness of the myocardial mass of the interventricular septum and the inferior-posterior and lateral walls (**Figure 1**), with the presence of trabeculae at that level and multiple recesses, of spongiform appearance, with flow inside (color Doppler, **Figure 2**). The noncompacted/compacted myocardial ratio was approximately 2:1.

There were neither alterations in the size of the

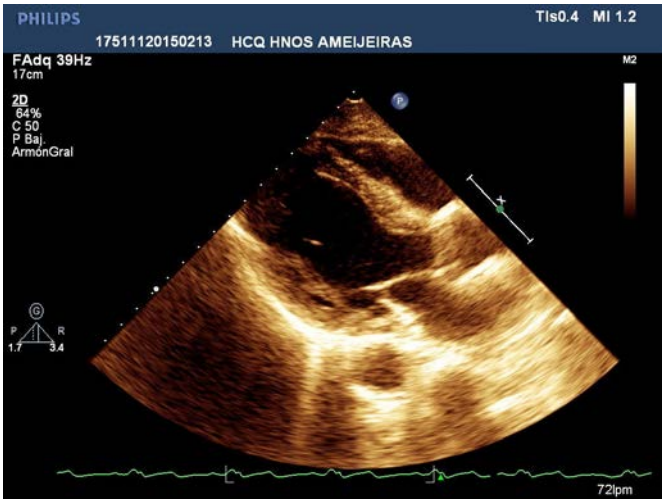

Figure 1. Parasternal long axis view where the increased myocardial mass is confirmed.

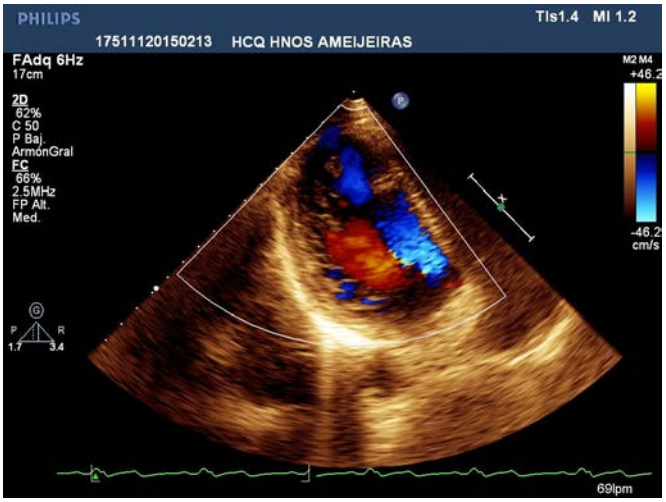

Figure 2. Endocardial trabeculae recesses demonstrated with color Doppler.

cardiac chambers or in the valvular apparatus, nor pericardial effusion, intracardiac thrombus or obstruction to the left ventricular (LV) outflow tract, which presented preserved segmental and global contractility, and a prolonged filling pattern; the right ventricle (RV) was of normal morphology and function.

With these elements, it was concluded as an isolated LVN and an automatic cardioverter-defibrillator (ACD) was implanted, after which the patient evolved favorably.

COMMENT

Syncope, transient loss of consciousness associated with muscle hypotonia that impedes the maintenance of normal postural tone, responds to numerous causes; i.e. it is necessary to direct correctly the diagnosis by an appropriate anamnesis[6].

The first diagnostic hypothesis in this patient was cardiogenic syncope, due to real loss of consciousness, short duration, with rapid recovery; in addition, it had been related to the efforts, which usually happens when it is cardiogenic by structural heart disease, and a murmur, characteristic of obstructive cardiomyopathy, was found. However, the episodes of syncope were also presented at rest[6], and there was no evidence of obstruction of the LV outflow tract in the echocardiogram –despite the diagnosis of spongiform cardiomyopathy– which decreased the possibility that they were caused by accompanying structural alterations. Nevertheless, with this diagnosis and the history of syncope, which sometimes appeared at rest, arrhythmias were considered as a causative agent, despite the absence of palpitations, but with evidence of a heart disease that is frequently associated to episodes of cardiac arrhythmias[7]. Although no arrhythmia was documented during the episodes of syncope, or at the admission, the existence of a left bundle branch block, an electrocardiographic abnormality, was demonstrated, which is recommended by the Clinical Practice Guidelines on the conduct for syncope[8], for suspecting the arrhythmic cause; which was the reason for implanting an ACD[8].

The neurally mediated syncope (reflex), either vasovagal, situational or by stimulation of the carotid sinus, was ruled out because the episodes were not triggered by emotional stress, stimulation of the sinus, or other specific scenarios such as coughing, sneezing, urination, swallowing or defecation, among others[6]. Also, the neurological origin by convulsions or cerebrovascular damage was ruled out because in these cases, the episodes are longer, accompanied by involuntary movements or signs of neurological targeting, usually preceded by auras and they can leave sequels[6].

The orthostatic hypotension was not responsible either, because no cause of primary autonomic failure (e.g., pure autonomic failure, multiple systemic atrophy, Parkinson's disease with autonomic failure), or secondary (e.g., diabetic neuropathy or amyloid) was not found; as well as volume depletion (e.g., bleeding or diarrhea)[6].

The term “isolated lack of compaction” was exposed by Chin *et al.*[2] in 1990, when they described 8 patients with no other associated cardiac alterations. This shows that this is a relatively recent disease, which is evident by the lack of consensus even in its name, having a great variety of denominations such as cardiomyopathy by absence of compaction of the LV, LVN, spongiform cardiomyopathy or hypertrabeculation of the LV[5].

Technological advances implemented in echocardiography equipment, the use of contrast media and cardiac magnetic resonance imaging have allowed a clearer visualization of the LV's apical area[5] and they have facilitated the diagnosis.

LVN is a disease that affects men more frequently; nevertheless, in women it has occurred with a greater extension of hypertrabeculation. On the other hand, it has been shown to have a genetic substrate[5]. This cardiomyopathy consists in a very rare congenital anomaly of the myocardium’s morphology, with very deep intertrabecular recesses freely communicated with the ventricular cavity. Thus, the myocardium has two layers: a thin subepicardial (compacted), and another thick subendocardial and trabecular (noncompacted)[9]. It takes place as a result of an arrest of the normal process of compaction of the ventricular wall, that occurs between 5 and 8 weeks of intrauterine life, and it is characterized by the progressive disappearance of sinusoidal intertubular spaces of the embryonic myocardium, which are transformed into capillaries within the coronary circulation. It develops from the epicardium to the endocardium, from the base of the apex and septum to the side wall, which would explain the most common sites of the noncompacted myocardium[10].

By definition, it takes place in the absence of structural heart disease and there are other areas of

thickened myocardium, with two layers, and which can be hypokinetic[11].

To recognize that the LVN is an eminently familiar disease has led to the search for genetic causes, and it has been shown that it is a heterogeneous condition from this point of view, which explains the variability in its patterns of inheritance, morphology and associated alterations[12].

Although in recent years great strides have been made in understanding the genetic origins and biology of cardiomyopathies, their classification remains complex and controversial. As established by the World Health Organization (WHO)/International Society and Federation of Cardiology (ISFC)[13], the LVN would be an unclassified cardiomyopathy; however, it is already included as primary (i.e., predominantly affecting the myocardium), with a genetic base[1].

The clinic of LVN can range from asymptomatic patients to severe heart failure with depressed LV systolic function, systemic embolism, arrhythmias (atrial fibrillation to sustained ventricular tachycardia), and conduction disorders such as the left bundle branch block or sudden death[5].

It should be considered when a patient meets at least two of the following three criteria: a) heart failure without apparent cause; b) suggestive echocardiogram, and c) systemic suggestive manifestations of LVN[5].

The electrocardiogram is usually abnormal, with nonspecific repolarization alterations, bundle branch blocks, left predominance, and atrial arrhythmias, ventricular, or both[14]. The echocardiography is essential for diagnosis (class I recommendation), and it is considered the standard method. Although there are no unified criteria, the most used are shown in the **table** below. It is necessary to fulfill the last three to establish the diagnosis. The parasternal short axis view is the one that offers a better definition[11], and the affected segments are typically hypokinetic in symptomatic patients and those with impaired LV systolic function[11].

Familiar and sporadic forms of the disease have been described. The first represent between 20-50% of cases[17-21].

The treatment for these patients is similar to that of other cardiomyopathies. LV dysfunction should be treated by appropriate medical treatment for that situation. Anticoagulation is recommended in patients with atrial fibrillation, LV dysfunction with ejection fraction <40%, or both; as well as in patients with previous thromboembolic events. The Holter monitoring is recommended once a year, to detect arrhythmias in asymptomatic patients, and indications for ACD implantation are similar to those for dilated cardiomyopathy. The option of heart transplantation should be taken into consideration when the intensive standard treatment for heart failure is not enough[22,23].

Table. Diagnostic criteria of Jenni *et al.*[11] for the LVN.

1. Absence of coexistent cardiac anomalies.
2. Marked thickening of the LV wall, divided into two layers:
A. Endocardial layer markedly thickened with numerous prominent trabeculae and deep recesses: noncompacted.
B. Thin epicardial layer: compacted.
• Relationship between the noncompacted and the compacted portions in tesystole > 2.
3. The endocardial recesses characteristically present detected flow with color Doppler.
4. Locations of noncompacted segments:
• Apical, middle or both segments: inferior and lateral wall of the LV.

The prognosis of these patients is unclear, although it is known that they evolve better when they are asymptomatic at the time of diagnosis[24]. In the first paper, which described a long-term follow-up (mean 44 months) of the symptomatic LVN[4], the 34 patients had a poor outcome, as 41% had ventricular arrhythmias; 35%, sudden death; 24%, thromboembolic episodes, and 12% needed heart transplantation. The clinical predictors associated with poor prognosis were: higher telediastolic diameters, worse functional class of the New York Heart Association [NYHA(III/IV)], chronic atrial fibrillation and bundle branch blocks[24]. Meanwhile, Murphy *et al.*[25] found a more favorable outcome in 45 patients, with a survival rate of 97% after a median follow-up of 46 months.

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