Noncompaction of the Myocardium in a Patient with Acute Myocardial Infarction

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Left-ventricular non-compaction (LVNC) is a rare congenital cardiopathy, which results from the failure of the myocardial compaction process, leading to the persistence of numerous and deep trabeculations communicating with the ventricular cavity. Its main clinical manifestations are heart failure, arrhythmias (supraventricular or ventricular) and episodes of arterial embolism.

The present case reports on a Brazilian patient living in Portugal, who was hospitalized due to acute myocardial infarction, which resulted in severe LV systolic dysfunction. During the patient's assessment, LVNC was diagnosed.

The clinical presentation is described, as well as the echocardiographic evaluation (two-dimensional and three-dimensional), nuclear magnetic resonance (NMR) imaging and ventriculography results. The diagnostic criteria and therapeutic options are discussed.

Introduction

Left-ventricular non-compaction (LVNC) is a rare congenital cardiopathy, which results from the failure of the myocardial compaction process between the fifth and the eight weeks of embryogenesis, leading to the persistence of numerous and deep trabeculations, communicating with the ventricular cavity[1]. Considering that the trabeculation occurs from the basis to the apical region and from the epicardium to the endocardium, the apical alteration is considered the predominant type. Its main clinical manifestations are heart failure, arrhythmias (supraventricular or ventricular ones) and episodes of arterial embolism[2]. An increasing number of cases have been observed in asymptomatic individuals.

Although described as rare until recently (with a prevalence in adults estimated at 0.014%)[3], recent studies have suggested that it is underdiagnosed: in a recently published study, almost 25% of the patients referred to a Heart Disease Clinic who presented LV systolic dysfunction met the criteria for this diagnosis[4].

Key words

Myocardial infarction; cardiomyopathies; cardiovascular diseases.

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Manuscript received March 22, 2009; revised manuscript received August 01, 2009; accepted October 08, 2009.

The diagnosis is achieved through echocardiography (according to the subsequently described criteria), with nuclear magnetic resonance (NMR) being reserved for diagnostic confirmation[5].

In series, up to 44% of the patients have a family history of cardiomyopathy. An X-linked autosomal dominant and mitochondrial transmission has been described in children. In adults, there is a predominance of autosomal dominant forms, whereas the X-linked forms are less frequent.

Mutations have been described in several genes, including tafazzin (TAZ/G4.5), LIM domain binding protein 3 (ZASP/LPB3), α-dystrobrevin (DTNA) and lamin A/C (LMNA). A recently published study identified gene variants encoding proteins of the sarcomere (MYH7, ACTC and TNNT2) in affected adult individuals. These same genes are associated with hypertrophic and dilated cardiomyopathy[6].

Case report

A 45-year-old Caucasian male individual from Brazil, who had been living in Portugal for three years, came to the Emergency service of our hospital due to cold sweats, dizziness, headache, vomiting after meals and periods of disorientation. He had no prior history of heart disease. His personal history included arterial hypertension, type 2 diabetes mellitus, hypercholesterolemia and he was a current smoker (smoking load of 60 pack-years). He had presented a prior episode of unstable angina four months before, when he was submitted to a cardiac catheterism and angioplasty with drug-eluting stent implantation in the mid-anterior descending artery (2.50*20mm). The two-dimension echocardiogram showed increased left ventricular (LV) dimensions, anterolateral hypokinesis and good global systolic function. The patient presented poor adherence to therapy and hygienic-dietetic recommendations, maintaining the habit of smoking and not taking clopidogrel.

The physical examination was normal at the second hospitalization; the 12-lead electrocardiogram showed anteroseptal fibrosis and ST-segment elevation in V4-V5, suggesting ST-segment elevation myocardial infarction (STEMI). He was submitted to emergency catheterism, which disclosed intra and pre-stent thrombosis in the mid-anterior descending artery, which was resolved using balloon angioplasty. The ventriculography showed severe LV systolic dysfunction with akinesis of the anterior wall and apex. The ejection fraction (EF) was 21% (in RAO). He was submitted to a two-dimensional transthoracic Doppler echocardiography and three-dimensional echocardiography. He presented dilation of

the left atrium (4.6 cm) and left ventricle (6.0 cm of diastolic diameter), with severe EF decrease (30% Simpson 4C and 2C); akinesis of the lower 2/3 of the septal, apical and apical-lateral regions. The LV lateral-apical region disclosed marked trabeculation that was more than 2-fold thicker than the compacted region and trabecular flow. The three-dimensional images show the non-compacted regions (Figure 1).

The review of the ventriculography disclosed images that were suggestive of noncompaction of the myocardium.

The magnetic resonance (MR) imaging confirmed the diagnosis of noncompaction of the myocardium located on the LV lateral wall and apex (Figure 2).

No mutations were found in the TAZ gene. The echocardiogram was repeated 40 days after the STEMI, with maintenance of the severe left ventricular function depression. An implantable cardioverter-defibrillator (ICD) was implanted in September 2007.

The patient is currently in NYHA functional class II, presents good therapeutic adherence and no need for hospitalizations.

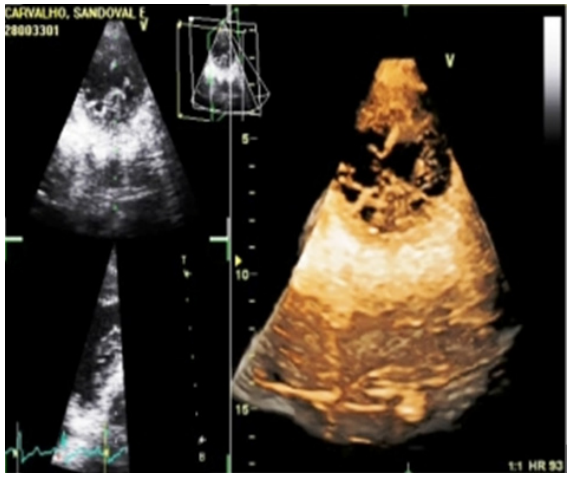

Figure 1 - Three-dimensional short axis view of the left ventricle (LV) disclosing marked trabeculations.

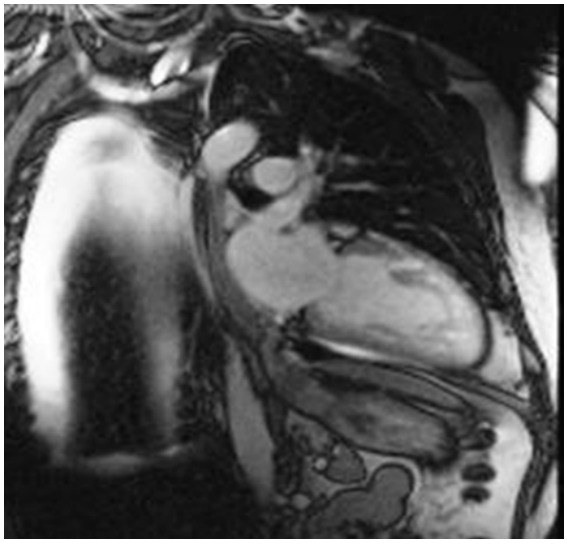

Figure 2 - Magnetic resonance imaging of noncompaction of the LV apical region.

Discussion

The first description of the "spongy myocardium" was made by Chin et al[4], in 1975. Since then, there has been much controversy regarding the criteria used for its diagnosis. Currently, the most widely accepted criteria are those established by Chin, Jenni and Stollberger's groups[7-9].

Chin's group defines it as a ratio < 0.5 between the distances: epicardium (start of the trabeculae) - epicardium (peak of the trabeculae). These must be located at the apex of the left ventricle, on the apical and the short parasternal axis view. The measurements correspond to the LV free wall thickness at the end of the diastole[7]. Jenni et al[5] maintains the need for the trabecular layer to be two-fold thicker than the non-trabecular layer and introduces the exclusion of coexisting structural cardiopathies, the concept of excessively prominent numerous trabeculations and deep intratrabecular recesses, with a Doppler blood flow originating from the ventricle[8].

Stollberger's criteria require more than three trabeculations protruding from the LV wall, distal to the papillary muscles and visible on the same plane; the intertrabecular spaces must be perfused from the ventricular cavity (seen at color Doppler imaging)[9].

Therapy is directed at the most frequent manifestations of the disease: heart failure treatment, prophylactic anticoagulation and pro-arrhythmic risk assessment. Due to the high prevalence of malignant arrhythmias, the Holter assessment must be carried out at least once a year. The electrophysiological study and/or the prophylactic implantation of ICD must be carefully considered. Competitive sports must be avoided and the first-degree relatives must be screened through Doppler echocardiographic studies[10].

The LVNC was initially described as having a poor prognosis: in a sample of 34 adult patients followed for 44 ± 39 months, 53% were hospitalized due to heart failure; 41% due to ventricular tachycardia and 24% due to embolic events; around 33% of the patients died and 12% received transplants[11]. However, more recent series have suggested a more favorable prognosis: one of these series showed a transplant-free survival of 97% at 46 months; another series of 65 patients showed that the individuals who were asymptomatic by the time of the diagnosis remained event-free at 43 months.

The patient described herein met the diagnostic criteria established by Chin et al[4]. Considering the presence of LVNC and the LV systolic dysfunction with an EF of 30%, the patient received an ICD and the medical therapy was optimized, with the patient currently being in NYHA functional class II. He has been advised to contact his first-degree relatives (who live in Brazil), so they can be submitted to a Doppler echocardiographic study to rule out the presence of LVNC.The present case illustrates the diversity of the disease presentation

and reinforces the need to search for characteristic trabeculations in the apical region in the presence of a dilated left ventricle with decreased function. It also reinforces the need for a broader consensus regarding the diagnostic criteria of this disease.

Potential Conflict of Interest

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

Sources of Funding

There were no external funding sources for this study.

Study Association

This study is not associated with any post-graduation program.

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