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Case Report

Isolated left ventricular non-compaction cardiomyopathy: case report and brief review on diagnostic imaging methods

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Abstract: Left ventricular non-compaction cardiomyopathy (LVNC) is a rare congenital heart disease with an estimated incidence of 0.014-0.045% in a general population. It occurs due to a morphogenetic abnormality during embryogenesis that inhibits myocardial compaction and determines prominent trabeculae with deep intertrabecular spaces and thickening of the adjacent myocardium in two distinct layers. Diagnosis is generally established via transthoracic echocardiogram (TTE) with further confirmation with cardiac magnetic resonance (CMR). A 57-year-old male patient previously diagnosed with systemic arterial hypertension and chronic obstructive pulmonary disease reported chest pain and dyspnea upon exertion, headache, and dizziness. During outpatient investigation, the patient was submitted to a stress electrocardiogram that was positive for ischemia and led to a cineangiocoronariography with no signs of coronary artery obstruction. At the echocardiography lab, several trabeculations were detected in the apical region of the left ventricle. A diagnosis of LVNC was established and the results were confirmed via CMR. Management was initiated with the goal of symptom relief. LVNC is a relatively unknown pathology that is frequently underdiagnosed at earlier stages. As presented, we highlight the importance of multimodality imaging, especially TTE and CMR, in the diagnosis of this congenital heart condition.

Keywords: Isolated Noncompaction of the Ventricular Myocardium; Echocardiography; Magnetic Resonance Imaging.

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1. Introduction

Left ventricular non-compaction cardiomyopathy (LVNC) is a rare congenital heart disease that is frequently under diagnosed due to its unspecific clinical presentation. It has an estimated incidence of 0.014-0.045% [1] in a general population (8-12 per million adults worldwide), accounting for the third cardiomyopathy most frequently diagnosed after dilated and hypertrophic cardiomyopathies [2, 3]. The prevalence of LVNC in cardiac patient cohorts diagnosed by echocardiography is estimated in 0.9%, whereas in patients diagnosed with cardiac magnetic resonance the number rises significantly to 9.6% [4-7].

The condition was first described in 1990 by Chin et al as "*isolated noncompaction of the left ventricular myocardium (INVM)*" [8]. It occurs due to a morphogenetic abnormality during embryogenesis that inhibits myocardial compaction and determines prominent trabeculae with deep intertrabecular spaces and thickening of the adjacent myocardium in two distinct layers: compacted and non-compacted with a ratio > 2:1 among them. The

term "isolated LVNC" can be used for areas of hypertrabeculation and normal left ventricular function with no evidence of either congenital or structural heart defects [9].

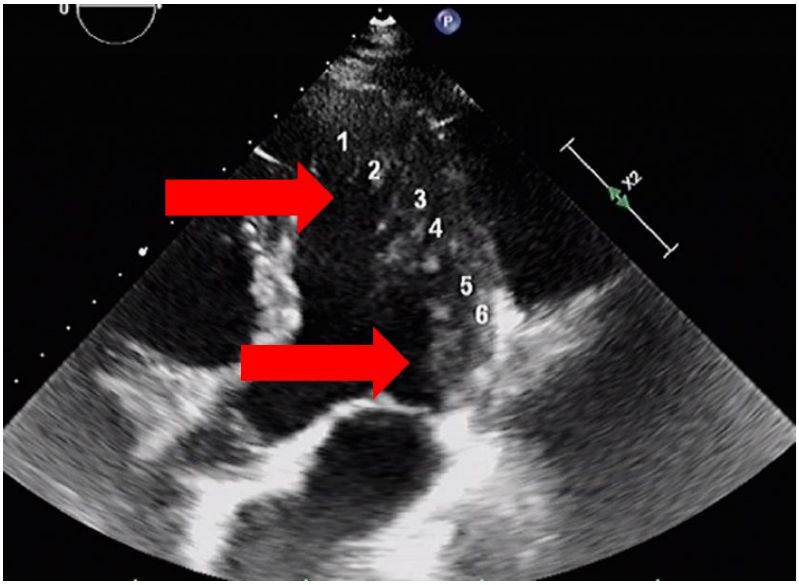
Although variable due to concomitant phenotypic variants, the main clinical presentation includes severe ventricular dysfunction, arrythmias, systemic thromboembolism and sudden cardiac death. There is also a documentation of LVNC phenotypes in health athletes and pregnant women [10,11], which suggests a physiologic adaptation in these specific patient populations with no significant clinical outcomes. Diagnosis is generally established via transthoracic echocardiogram (TTE) with further confirmation with other imaging methods, particularly cardiac magnetic resonance (CMR). Treatment of this condition is symptom-oriented and directed to the clinical presentation.

Therefore, the aim of this case report and brief literature review is to describe a relatively uncommon disorder and its multivariate clinical presentation. The uniqueness of isolated LVNC and the emerging role of imaging modalities in the diagnosis should raises concern about the disorder and stimulate further research in the field.

2. Case Report

A 57-year-old male patient with previous medical history of systemic arterial hypertension and chronic obstructive pulmonary disease (COPD) presented for further investigation of his ongoing cardiovascular condition. The patient reported progressive chest pain and dyspnea upon exertion that started 6 months prior to the current presentation. Accompanying symptoms also included headache and dizziness. During initial investigation, he was submitted to a stress electrocardiogram, which was positive for myocardial ischemia. Given the results of the non-invasive method, the cardiology team decided to pursue additional testing with cineangiocoronatiography. There were no signs of coronary artery obstruction, and the patient was referred to the echocardiography laboratory. Left ventricular functional was unremarkable and wall motion abnormalities were absent, but an unexpected finding was observed: hypertrabeculations in the apical region of the left ventricle. These findings were consistent with isolated LVNC (Figure 1).

Figure 1. Prominent trabeculae with deep recesses are evident on the following segments: lateral-medial and lateral-apical (Arrows in red and numbers 1-6).



The Color Doppler also demonstrated evidence of flow inside the trabeculae regions (Figure 2). A newer technique called Global Longitudinal Strain (GLS), which is derived from the bidimensional left ventricle speckle tracking was implemented. The results were

consistent with reduced GLS (-14.3% vs normal reference level of ≤ -18%) and segmental abnormalities on inferior and inferior-septal walls were present (Figure 3).

Figure 2. Evidence of flow by the Color Doppler inside the intertrabecular spaces (Indicated by numbers 1-3 and red arrows).

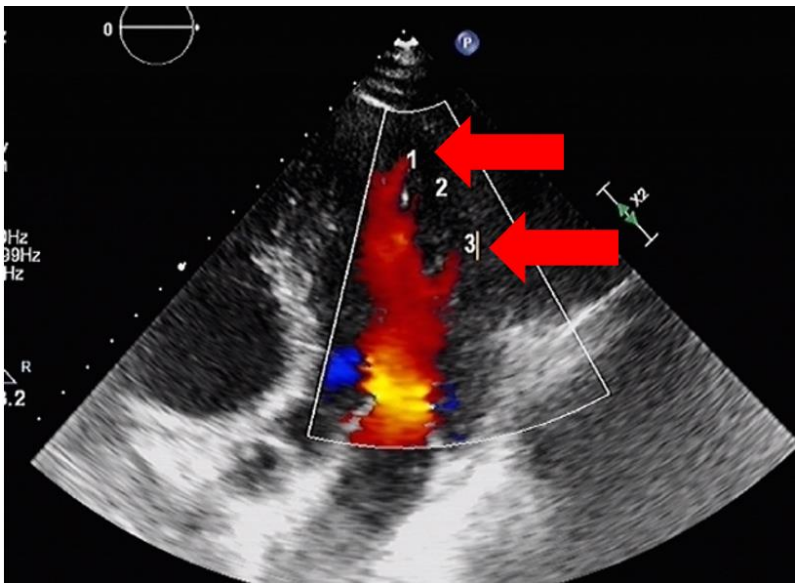
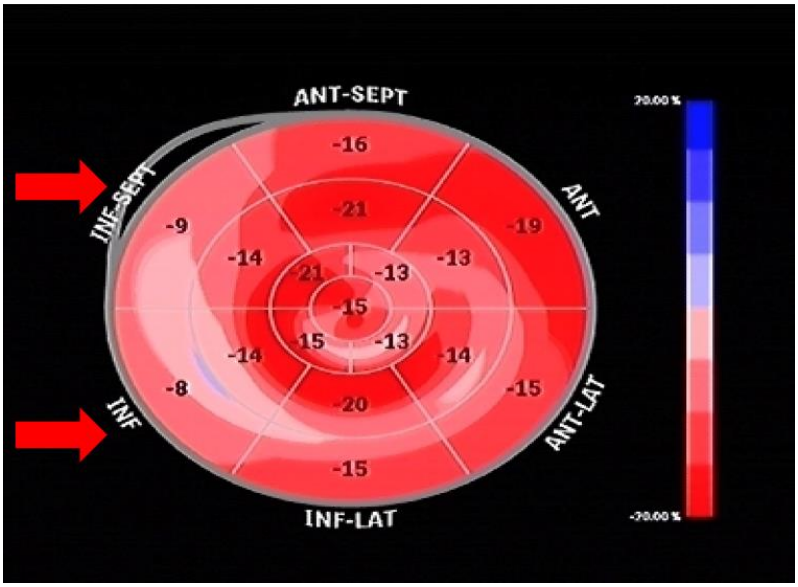
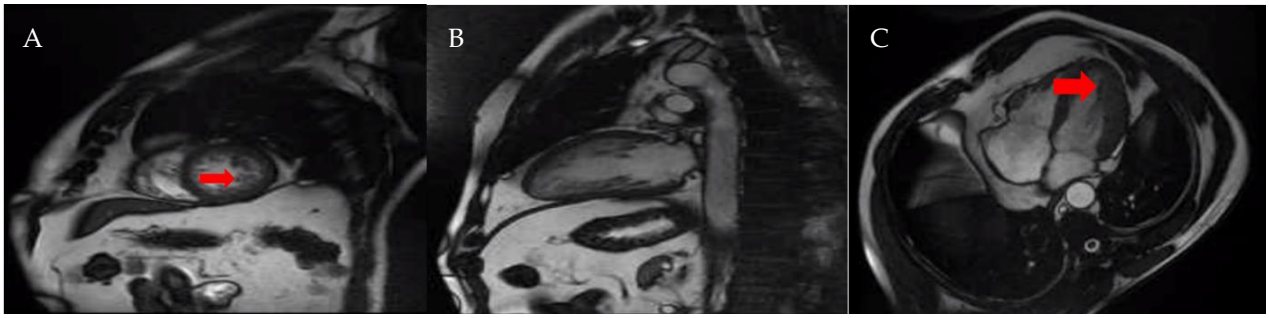


Figure 3. Bull's eye demonstrating reduced left-ventricle Global Longitudinal Strain obtained by bidimensional speckle tracking. Measured value: - 14.3% (Reference value ≤ -18%). Segmental abnormalities were more pronounced on the inferior and inferior-septal walls, as indicated by the red arrows.



We decided to confirm the TTE findings with CMR and a definitive diagnosis was made (Figures 4A to 4C). Additional screening for arrhythmias was performed with resting electrocardiogram and 24-hour Holter monitoring: normal sinus rhythm, normal QRS complex duration, absence of heart blocks or conduction abnormalities, and no evidence of ectopic supraventricular or ventricular activity. Genetic testing was performed, and additional family screening indicated in this scenario.

Figure 4. Cardiac Magnetic Resonance imaging on apical short and long axis indicating myocardial hypertrabeculation with a non-compactation ratio of 3:2 (highlighted by the red arrows).



Until now, there is no specific guideline-directed treatment for LVNC. Initial management typically consists in symptom relief and control of previous medical conditions. In this particular case there was no clear indication for implantable cardiac defibrillator, specific antiarrhythmic treatment, and anticoagulation. Thus, treatment plan consisted in beta-blocker (Metoprolol Succinate 100mg/daily), optimal management of patient's previous hypertension and COPD, and genetic testing of first-degree relatives. As stipulated by current guidelines, follow-up intervals are guided by disease severity. In the case presented by our group, the patient was considered a low-risk spectrum due to excellent symptom improvement with beta-blocker therapy, absence of left ventricular dysfunction or conduction abnormalities and negative family history of SCD. Therefore, follow-up visits were scheduled in intervals of 6 months with exams guided at patients' presentation at the time or at physician's discretion. The patient remained in asymptomatic for over 2 years with no relapse of disease. The was an overall improvement of the perceived quality of life, as reported by the patient.

3. Discussion and conclusions

LVNC is a complex entity with heterogeneous clinical presentation based on phenotype-specific abnormalities. It can be isolates (as reported in the case presented by our group) or associated with other heart conditions, particularly congenital heart diseases. The multivariate presentation puts at stake whether it is a true cardiomyopathy or a physiologic pattern. It is divided in benign, associated with arrhythmias, dilated, hypertrophic, dilated-hypertrophic, restrictive, and biventricular or right ventricular LVNC [12].

In the isolated variant, Jenni et al in agreement with morphologic necropsy findings published the most frequently used diagnostic criteria: ratio of non-compacted layer to compacted myocardium > 2 at the maximal end-systole [13]. Cardiac Magnetic Resonance has been rising as the next-step modality after TTE for the differential diagnosis of cardiomyopathies and LVNC. The most commonly alternate diagnosis include healthy individuals, left ventricular hypertrophy, thrombus, false tendons, endomyocardial fibrosis and eosinophilic heart disease [14]. Diagnostic criteria using CMR for left ventricle hypertrabeculation has been proposed by Peterson et al in 2005. These criteria evaluated the non-compacted to compacted layer ratio ≥ 2.3 measured in long axis cine views at the site with the most pronounced trabeculations [15].

In terms of medical management, there are no specific guidelines or treatment for this condition. The current understanding is symptom-oriented management based on the specific phenotypic presentation. One of the main topics is thromboembolic risk prevention: it is suggested that patients with left ventricle dysfunction (left ventricular ejection fraction < 40%) are at increased risk of systemic embolism and therefore may require oral anticoagulation [16]. Implantable cardiac defibrillator implantation is generally limited for patients with LVNC presenting with syncope or if other guideline-oriented conditions are present [17]. In view of the above, we reported a unique case presentation of a

commonly underrepresented condition with heterogeneous presentation and controversies regarding diagnostic criteria and even its existence as a true cardiomyopathy or as a physiologic variant. Therefore, further research is stimulated in order to establish definitive guideline-specific diagnostic, management criteria, and improve awareness about this disorder.

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Research Ethics Committee Approval: We declare that the patient approved the study by signing an informed consent form and the study followed the ethical guidelines established by the Declaration of Helsinki.

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Conflicts of Interest: None.

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