ISSN: 2577 - 8005

Literature Review

Medical & Clinical Research

Left ventricular non-compaction: Underdiagnosed disease with fatal outcomes

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Submitted:07 Feb 2023;**Accepted**: 20 Feb 2023;**Published**:07 Mar 2023

Citation: *Neelanjana Pandey, Miguel A Rodriguez Guerra, Maria J Duran, Harpreet Kaur, Timothy J Vittorio (2023) Left ventricular non-compaction: Underdiagnosed disease with fatal outcomes. Medical & Clinical Research 8(3): 01-03.*

Abstract

Left ventricular non compaction (LVNC) is a type of congenital cardiomyopathy characterized by the presence of prominent intertrabecular recesses that typically affect the apical portion of the left ventricular cavity. These recesses give a distinctive phenotype to the affected myocardium that has been described as "sponge-like". Devastating complications have been observed in those patients with LVNC, including heart failure requiring heart transplantation in selected cases, life-threatening arrhythmias, thromboembolic events including stroke and transient ischemic attack and sudden cardiac death. This report follows the case of a 62-year-old female patient with recurrent chest pain for several years and extensive negative work and recent echocardiography showed increased left ventricle trabeculations, suspicious for LVNC, later confirmed by cardiac magnetic resonance (CMR).

Keywords: Sudden Cardiac Death, Cardiac Magnetic Resonance, Cardiomyopathy, Heart Failure, Tachycardia

Introduction

LVNC affecting left ventricular myocardium is a relatively rare and under recognized cause of cardiomyopathy with potentially fatal consequences. It was first described in 1984, the estimated prevalence of this congenital cardiomyopathy is estimated to be at 0.05% [1] in the general population and potential outcomes include cardiac failure, thromboembolism, and malignant arrhythmias. A study of 960 heart failure patients identified LVNC in 3% as the etiology of heart failure [2], notably the same prevalence as hypertensive heart disease in that same center.

A systematic review found that patients with LVNC had 8.6% increase of major cardiovascular events such as cerebrovascular accident and death for the time period that they were observed [3]. The study also found increased frequency of sustained and non-sustained ventricular tachycardia and atrial fibrillation in patients with LVNC.

Another study suggests that the risk for thromboembolic events is markedly elevated in these patients such that there is sufficient evidence to consider anticoagulation especially in the setting of previous thromboembolic events or left ventricular systolic impairment, but this is yet to be more investigated. Despite all these findings, the long-term outcomes are not well known for those affected by this condition, hence more research and awareness are of vital importance.

Case Presentation

This is a case of a 62-year-old female who presented to the general cardiology service with a worsening mid-sternal chest pain, described as dull and constant for one week, it is radiated to her left breast and her left arm. This pain was the same pain that had been going on for 8 years and occurred almost daily. For the last week, the pain had become constant, non-exertional, occurring at rest, lasting several minutes and was not associated with any diaphoresis, palpitations or shortness of breath. She has a medical history of hypertension, bronchial asthma, dyslipidemia, and chronic atypical chest pain. Patient had no family history of cardiovascular disease including sudden cardiac death, denied smoking or alcohol consumption.

Prior work up showed patent coronary arteries in a cardiac catheterization a year prior.

At the emergency department electrocardiogram revealed normal sinus rhythm and T wave inversion in leads V3, V4, V5. Serial measurements of cardiac enzymes were negative. Computed Tomography of the chest with contrast showed no active cardiopulmonary pathology.

The echocardiogram revealed increased left ventricular trabeculations suggestive of LVNC (Figure 1). A cardiac magnetic resonance showed apical and anterolateral left ventricular walls

trabeculations suggestive of ventricular noncompaction, with a 3:1 ratio, compatible with diagnosis of LVNC. Patient and family were educated, subsequently, the first-degree family members were screened and two of her children were diagnosed with LVNC.

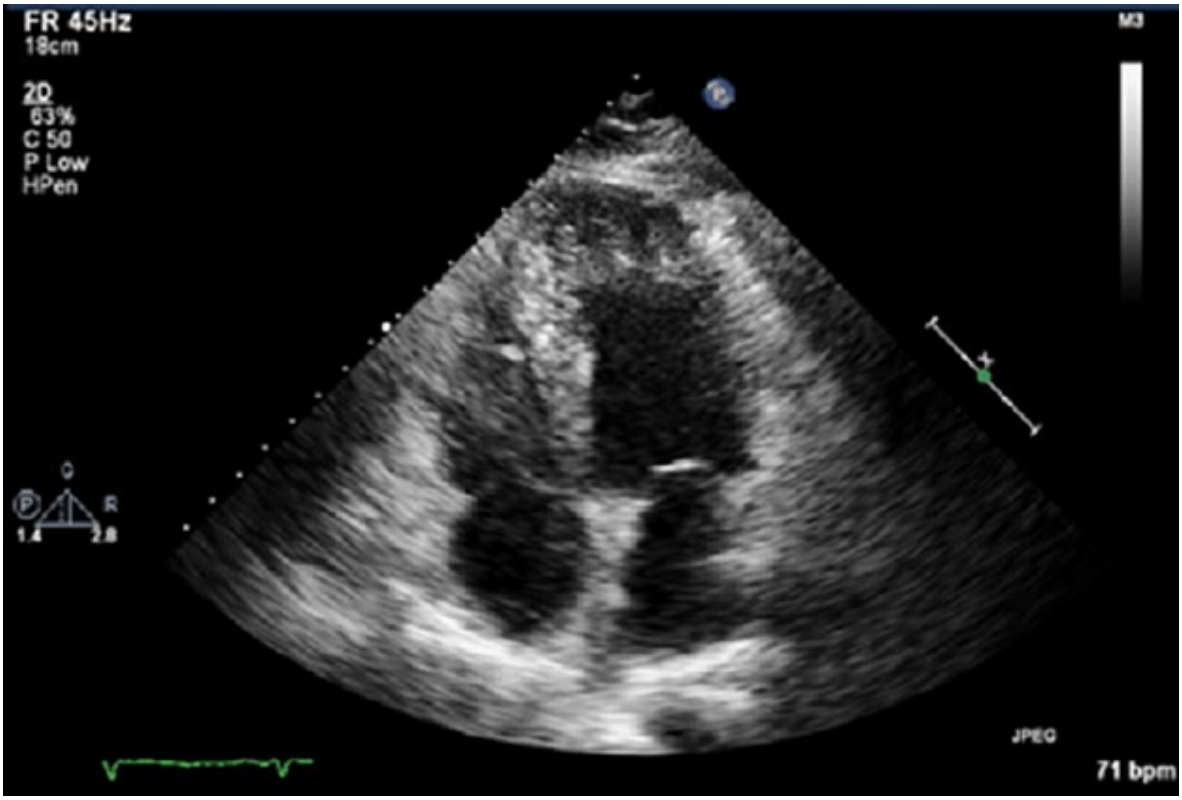

Figure 1: Apical four chamber view in TTE contrast revealed prominent apical trabeculations (arrow) suggestive of ventricular noncompaction.

Currently, the patient continues to experience paroxysmal episodes of atypical chest pain but has not developed palpitation, dyspnea, syncope or extremity swelling. She continues to follow up in the cardiology clinic where the left ventricular function is assessed periodically. The left ventricular ejection fraction has been preserved on serial echocardiograms. Her current medication regimen comprises aspirin, nifedipine and carvedilol. Patient is also periodically screened for arrhythmias with Holter monitor.

Discussion

Left ventricular non-compaction (LVNC) is a relatively uncommon cause of cardiomyopathy [4]. Despite not having a gold standard diagnostic criteria, this condition can be recognized when certain morphologic findings are present using echocardiography and cardiac magnetic resonance (CMR) [5]. These include presence of prominent intertrabecular recesses affecting the left ventricle myocardium, along with blood flow from the ventricular cavity to the recesses [6]. To make the diagnosis there should be no communication between the blood that flows from the ventricular cavity to the recesses and the coronary circulation [7]. On histopathologic examination, findings commonly include prominent trabeculae surrounded by deep intertrabecular recesses covered by ventricular endocardium. Ischemic lesions and variable degree of interstitial fibrosis are frequently observed [8]. It is possible to identify severe disease using the forementioned morphologic criteria however milder phenotypes have been described, sometimes with overlap with physiologic changes. In these settings CMR allows for better assessment of structure and presence of fibrosis; if it is not performed, diagnosis could be controversial [9].

There is a higher incidence of thromboembolic events including stroke and transient ischemic attacks in patients with LVNC [10]. The risk of life-threatening arrhythmias and heart failure in patients is also increased, the therapy is based on the standard management or established guidelines as in those patients without LVNC[11]; same principle applies to other complications such as life-threatening arrhythmias and thromboembolic events [12]. Due to these complications, early diagnosis is vital, and some studies advise screening first degree relatives with echocardiography and genetic studies [13].

A significant number of patients present with recurrent episodes of atypical chest pain, while others may complain of palpitation, dyspnea or syncope [14]. These symptoms can be overwhelming to patients and can lead to extensive and invasive medical workup to try to identify the etiology [15]. Usually the diagnosis is delayed, causing more distress to the patient and increased utilization of resources and healthcare cost [16].

The sensitivity and specificity of the current diagnostic criteria are not known, and it may suggest that the prevalence of LVNC and its role in the increased morbidity & mortality may be greater [17]. The current literature about the long-term effects of LVNC is limited [18]. The available literature about LVNC is limited, but its complications could lead to thromboembolic phenomenon, arrhythmias, and death [19]. Although multiple complications have been described, there is a need for further investigation regarding diagnosis, screening and management for this cardiomyopathy [20].

Conclusion

This is an exceptional case of left ventricle non-compacted cardiomyopathy of a patient where a hereditary familiar pattern was confirmed. This case exposes the importance of the genetic screening on this condition and how it would allow the early approach to the family members potentially impacting their outcome positively.

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