

Isolated left ventricular non-compaction: Clinical characterisation of a teenage male

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Abstract

Isolated Left Ventricular Non-compaction (LVNC) is a type of cardiomyopathy that usually has a genetic origin. Its diagnosis is based on finding such as deep intertrabecular recesses or sinusoids and ventricular trabeculations communicating with the left ventricular cavity. LVNC was first clinically recognised almost four decades ago, yet its diagnostic and management challenges persist. In this report, we present the case of an 18-year-old boy, who presented at the National Institute of Cardiovascular Diseases, Karachi, in March 2023, with complaints of dizziness, pedal oedema, and shortness of breath. Echocardiography revealed signs suggestive of LVNC, which were confirmed conclusively on Cardiovascular Magnetic Resonance (CMR) (NC/C ratio > 2.4). The patient underwent implantable cardioverter defibrillator (ICD) placement, was discharged after a smooth post-procedure recovery, and is doing well on follow-ups. Hence, ICD and guideline-directed medical therapy as a combination have turned out to have satisfactory outcomes in decreasing morbidity and providing mortality benefits for such patients.

Keywords: Left ventricular non-compaction, Isolated non-compaction of left ventricular myocardium, Left ventricular dysfunction.

DOI: <https://doi.org/10.47391/JPMA.10034>

Introduction

Isolated Left Ventricular Non-compaction (LVNC) is a cardiomyopathy of genetic origin characterised by the presence of deep intertrabecular recesses or sinusoids and ventricular trabeculations in communication with the cavity of the left ventricle.¹ It is also defined as a type of myocardium that consists of two layers, non-compacted (NC) and compacted (C).² LVNC remained an unclassified type of cardiomyopathy until 2006, when it was classified by the American Heart Association as primary

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Submission complete: 25-05-2023

Review began: 29-06-2023

Acceptance: 13-03-2024

Review end: 24-12-2023

cardiomyopathy with genetic predisposition.³ To date, it stands as a separate entity among cardiomyopathies that has scarce literature and stays under-investigated most of the time.

The pathological cause of LVNC dates back to the first trimester when non-compacted myocardium can be visualised by trabeculations originating from the endocardium. By the end of the first trimester, this layer of the myocardium with trabeculations undergoes compaction, hence, causing the majority of the myocardial volume to be compacted. The process of compaction continues postnatal. Any pathology that delays or puts a halt to compaction may hamper the development of spiral fibres of the myocardium which is the final process responsible for the efficient rotational ventricular contraction system. This can, in turn, cause myocardial dysfunction secondary to the loss of the twisting nature of ventricular contraction.⁴ The overall familial recurrence of this disease was noted to be 30% by Bhatia et al,⁵ however, another study concluded that 60-70% of the cases had sporadic occurrence.⁶

Even after several research studies, the pattern of occurrence, diagnosis, and management of LVNC remains undetermined. Hence, it is of utmost importance that such cases be documented, highlighting the familial predisposition, diagnostic modalities used, and selected approach for increasing the literature pool of LVNC. This case report aims to highlight the presentation, course of actions, and outcomes of this rare case of Isolated LVNC.

Case Report

After obtaining consent from the patient, presented here is the case of an 18-year-old boy, who came to the outpatient department of National Institute of Cardiovascular Diseases Karachi in March 2023, with complaints of dizziness for one year, which usually led him to fall while standing, without losing consciousness. The last reported episode of a fall secondary to feeling dizzy was almost a month back. He also complained of noticing facial puffiness and pedal oedema along with shortness of breath on minor exertion (functional class III) for the last three months. The patient did not have any known comorbidity or prior medical or surgical history. His family history was also non-significant for any familial heart

conditions.

On examination, the patient was a healthy teenage boy with a Body Mass Index (BMI) of 20.6 kg/m². His blood pressure averaged at 106/54 mmHg with a pulse rate of 58 bpm. Pulse was of normal volume, normal character, and regular rhythm. On inspection of the precordium, there were visible pulsations at the left sixth intercostal space (ICS) lateral to the midclavicular line. There was no visible chest deformity or scars. On palpation, no thrill or parasternal heave was noted. The apex beat was displaced (palpable at left 6th ICS in anterior axillary line). The character of the apex beat was noted to be forceful and ill-sustained. On auscultation of the precordium, both S1 and S2 heart sounds were audible. S1 heart sound was normal in character while P2 was loud, and a pan systolic murmur of grade 3/6 at the left 3rd and 4th ICS near the left parasternal border (tricuspid area) was also appreciated. On auscultating the chest, normal vesicular breathing was appreciated. Jugular venous pulse (JVP) was normal and the rest of the general physical examination was unremarkable.

The electrocardiogram (ECG or EKG) displayed bradycardia, QRS duration 0.16s, v-voltage signs suggestive of left ventricular hypertrophy with a strain pattern and inverted T-waves in V2-V4 and aVL. To further help in the process of diagnosis, a 2-Dimensional (2-D) echocardiogram was performed. The echo showed biatrial and biventricular dilatation. The left ventricle (LV) showed multiple intertrabecular recesses communicating with the left ventricular cavity, suggestive of the diagnosis of LV non-

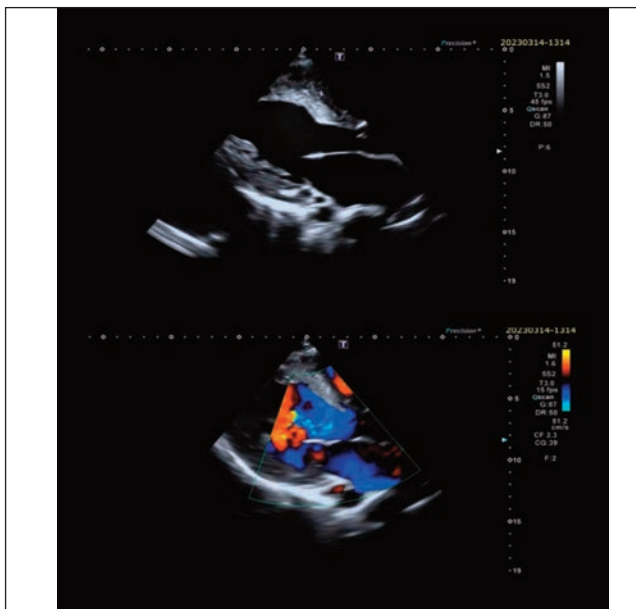


Figure-1: Echocardiogram showing Left Ventricular Non-compaction (LVNC).
(Multiple trabeculations with deep intertrabecular recesses in communication with the cavity of the left ventricle can be seen.)

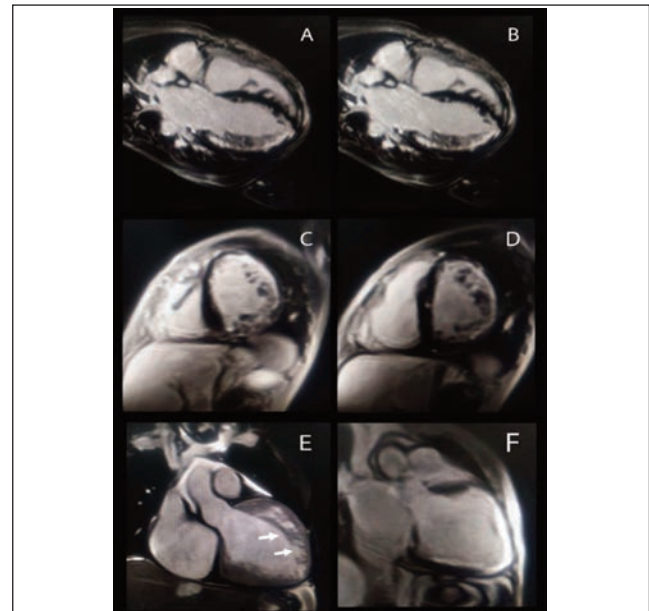


Figure-2: CMR showing Left Ventricular Non-Compaction.
Images A and B are showing 4-chamber views, C and D are showing short-axis views and E and F are showing parasternal long-axis views. Image C shows Late Gadolinium Enhancement (LGE) while Image E shows multiple coarse trabeculations with deep intertrabecular recesses forming the non-compacted layer of Left ventricle (arrows).

compaction (LVNC) (Figure 1). Valvular reports included severe tricuspid regurgitation, moderate pulmonary regurgitation, moderate mitral regurgitation, and trace aortic regurgitation. It also revealed moderate pulmonary hypertension and a TAPSE of 25mm. The inferior vena cava was dilated and non-collapsing. Flow reversal was seen in the hepatic vein.

To confirm the diagnosis, Cardiovascular Magnetic Resonance (CMR) was done (Figure 2). It revealed dilated LV with mild LV systolic dysfunction and coarse trabeculations of the anterior interventricular septum (IVS), anterior and lateral walls, and LV apex. Non-compacted to compacted myocardium ratio (NC/C ratio) at the end diastole was >2.4. An accessory muscle band was also noted in LV. Early gadolinium images showed no areas of hypo enhancement to suggest intracavitary thrombus. Late gadolinium enhancement (LGE) images showed mild myocardial hyper enhancement of LV apex, anterior and lateral walls. Mild RV systolic dysfunction was also noted. Other values included LVEF 45%, LVEDV 353ml, LVEDVI 210ml/m², and LVESV 192 ml. The findings of CMR were diagnostic of LVNC.

Since there is no definitive treatment for LVNC, our approach was to manage the patient symptomatically. It was decided to place an implantable cardioverter defibrillator (ICD) as a part of the patient's treatment for mortality benefit. The patient underwent the procedure

successfully and was discharged on guideline-directed medical therapy.

Discussion

Left Ventricular Non-compaction (LVNC) is an uncommon disorder with the genetic aberration of endocardial and myocardial morphogenesis, which got its first clinical recognition in 1984 by 2D echocardiography.⁷ Due to its unclear origin and under-investigated approach, the mortality rate is reportedly high (75% six-year mortality in symptomatic patients).⁸ LVNC may present as an isolated condition or associated with congenital heart disease. Medical archives have scarce data on the management and approach towards isolated LVNC, and most of these cases were diagnosed at autopsy.⁹

A study documented that LVNC can be diagnosed anywhere between birth and adolescence like in this case, the patient presented at 18 years of age while his symptoms developed after he turned 17.¹⁰ This patient's complaint of pedal oedema and dyspnoea was supported by a case of a 29-year-old, reported in 2021, who presented with similar symptoms at the time of their diagnosis; however the complaint of dizziness was not common among other reported cases.¹¹ Our patient presented with symptoms that were because of heart failure secondary to reduced ejection fraction only, one of the three major clinical manifestations of LVNC forming a triad of ventricular arrhythmias, heart failure, and systemic embolic events.¹¹

Echocardiogram was used as a first investigation of choice, which has been accredited as the most commonly used initial test by Bennett CE et al⁴ in their literature review that may lead to further evaluation by highlighting a few characteristic findings of LVNC. The case by Toader D et al¹¹ also highlighted reduced ejection fraction, presence of trabeculations, multiple apical thrombi between trabeculations, and moderate mitral regurgitation on echocardiography in their case, whereas in the present case only reduced ejection fraction and moderate mitral regurgitation were discovered.

A definitive diagnosis in this case was made on the findings of Cardiac Magnetic Resonance (CMR), such as the Non-Compaction to Compaction ratio (NC/C ratio) in diastole and Late Gadolinium Enhancement (LGE).^{12,13} The criteria for diagnosing LVNC based on CMR was described by Petersen et al¹² in 2005, which states that an NC/C ratio of >2.3 in diastole can distinguish pathological LV non-compaction, with 86% sensitivity and 99% specificity. This criterion stood accurate in the diagnosis of the present case where the NC/C ratio was >2.4. Dodd et al¹³ in their study compared the extent and severity of non-compaction and

the degree of late gadolinium enhancement (LGE). Results concluded that the severity of delayed enhancement varied among patients as mild, moderate, and severe, which correlates with the current patient's findings of mild myocardial hyper enhancement of LV apex, anterior and lateral walls on LGE images.¹³

In such patients, placement of implantable cardioverter defibrillators is still a dilemma in the line of primary prevention from sudden cardiac death (SCD). As per Bennett CE et al,⁴ ICD placement is reasonable if a patient with reduced EF meets the criteria for its sustained ventricular tachycardia, or history of SCD in the family, then the clinicians must weigh ICD's risk as compared to benefits for that particular patient to make any decision. The current patient had a reduced EF without any of the above-mentioned factors, hence he underwent ICD placement and had a smooth post-procedural recovery.

As defined by Bennett et al,⁴ there are two categories that lie in absolute indication for anticoagulation in patients with LVNC: 1) LV clot/thrombus visualised on imaging, or 2) documented atrial fibrillation. Patients who do not fall in either of these categories must be given anticoagulation only after their risk assessment is done via CHADS2-Vasc score. As far as the treatment/management after discharge of this patient is concerned, he was given drugs for heart failure only as he did not qualify for anticoagulation after his CHADS2-Vasc score was calculated.

Conclusion

After almost four decades since its first clinical recognition, the diagnostic and management challenges for Left ventricular non-compaction (LVNC) persist. Based on our case's clinical evidence, echocardiography is still credible as the most common initial test that may lead to further evaluation by highlighting characteristic findings of LVNC. CMR stood as a gold standard investigation in providing an NC/C ratio for definitive diagnosis. ICD and guideline-directed medical therapy as a combination can help in decreasing morbidity and adds mortality benefit in patients with LVNC.

Disclaimer: None.

Conflict of Interest: None.

Funding Sources: None.

Abbreviations:

LVNC: Left Ventricular Non-compaction

CMR: Cardiac Magnetic Resonance

NC/C ratio: Non-compaction to compaction ratio

IVS: Interventricular septum

ICS: Intercostal space

ICD: Implantable cardioverter defibrillator
 BMI: Body mass index

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Author Contribution:

JK: Conceived idea, design and responsible for the integrity of research.
 HS, PA: Review and final approval.
 AQM, BJ: Editing.