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Case Report

A Case of Pregnant Woman with Severe Mitral Regurgitation Demonstrating Left Ventricular Non-compaction

Alekhya Abburu[1*], Rahul Reddy Karnati[2]

[1]Resident Doctor, Department of Cardiology, Care Hospitals, Banjara Hills, Hyderabad, India
[2]DNB Resident, Department of Cardiology, Care Hospitals, Banjara Hills, Hyderabad, India
***Correspondence author:** Alekhya Abburu, Resident Doctor, Department of Cardiology, Care Hospitals, Banjara Hills, Hyderabad, India;
Email: dralekhya97@gmail.com

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Abstract

Left ventricular non-compaction is a rare form of cardiomyopathy. It is characterized by a thin compacted epicardial layer and a noncompacted endocardial layer with significant wall trabeculations and intertrabecular recesses that communicate with the ventricular cavity. It could be either in isolated form or coexists with other congenital heart diseases including valvular heart disease. It is associated with congestive heart failure, life threatening cardiac arrhythmia and thromboembolic complications. Adult LVNC has a prevalence of 0.01-0.27%. Diagnostic workup includes echocardiography, contrast ventriculography, and CT/MRI. Cardiac magnetic resonance imaging provides greater sensitivity and specificity, with a noncompacted to compacted myocardium ratio of > 2.3. Here we present an isolated case of Left ventricular non-compaction in a 21-year-old female due to underlying heart failure, suspected of Chronic Rheumatic Heart Disease. There is no specific treatment for LVNC, therapeutic measures are used for patient symptom relief with consideration of implantable cardioverter defibrillator and cardiac transplantation.

Keywords: Left Ventricular Non-Compaction; Rheumatic Heart Disease; Intertrabecular Recesses; Cardiac MRI; Echocardiography; Implantable Cardioverter Defibrillator (ICD); Cardiac Transplantation

Introduction

One of the rarest forms of genetic cardiomyopathy diagnosed so far is the Left Ventricular Non-Compaction (LVNC) It is characterized by deep intertrabecular recesses and aberrant trabeculations which occur due to failure of phase of cardiac development and thickening of myocardium into two distinct layers- compacted and non-compacted [1]. Though the recent research suggests the etiology as failure of developmental embryogenesis, the exact pathophysiology of the condition still remains controversial [2]. The most frequently involved areas include left ventricular apex, inferior and lateral walls and in rare cases right ventricular apex may also be involved [3]. The American Heart Association classified it as a primary genetic cardiomyopathy. A recent analysis, revealed that 18-42% of non-compaction cases were familial [4]. Historically LVNC has been diagnosed by echocardiography, which helps visualize apical region optimally leading to underestimation of degree of LV non compaction. However, Cardiac MRI provides a comprehensive depiction of cardiac morphology, suggesting the precise ratio of noncompacted to compacted myocardium of >2.3 [5]. We now present a case of a young female with isolated LVNC diagnosed using cardiac MRI.

Ethical Statement

The project did not meet the definition of human subject research under the purview of the IRB according to federal regulations and therefore was exempt.

Epidemiology and Genetic Studies

LVNC is a rare disease with a prevalence of 0.01-0.3% in adult population, demonstrating higher male predominance with 56% more than female [6]. The criteria for diagnosis are not uniformly defined and depend on the underlying disease entities such as congestive heart failure, cardiac arrhythmias and thromboembolism. LVNC exists in both sporadic and familial forms. It usually occurs as Autosomal dominant, but can be seen in X-linked recessive and mitochondrial subtypes [7]. LVNC is common in genetic cases with genes MYBPC3, MYH7, and TTN commonly involved [8]. This disease entity can be found in association with several cardiomyopathies including DCM, HCM and neuromuscular and metabolic diseases [9]. However, Heart failure remains the most common underlying cause. Patients with sporadic form have good prognosis and better recovery outcomes.

Diagnostic Approach

The gold standard for diagnosis of LVNC is echocardiography, nonetheless, there are alternate treatment modalities such as Cardiac MRI. Area of non-compaction can be better identified with CMR, as it allows for tissue evaluation and quantifies the extent of non-compacted myocardium [10]. The CMR criteria of Peterson, et al., for diagnosis are a non-compacted/compacted myocardium ratio > 2.3 in a long-axis end-diastolic image in at least two consecutive segments. Andreini, et al., identified LV dilatation, systolic dysfunction and late contrast enhancement as independent predictors of poor outcomes. In a retrospective study with a group of patients, it was observed that MR was commonly seen with LVNC in systolic dysfunction [11]. Here, in this case study, we evaluated a pregnant woman with severe LV dysfunction and severe mitral regurgitation known to have underlying LV non-compaction using Cardiac MRI as final diagnostic modality. LVNC remains a missed diagnosis till the present day, as there are no specific presenting signs and symptoms. Proper diagnostic and therapeutic regimen need to be followed for timely management. All suspected patients admitted with congestive heart failure, and ventricular arrhythmias must undergo thorough diagnostic workup with echocardiography followed by contrast-enhanced CMR. LVNC is not a fatal condition and has a better prognosis with proper management.

Case Presentation

A 21-year-old G2A1, known hypothyroid with a previous history of Spontaneous abortion at 16 weeks gestation, complaints of having severe shortness of breath, orthopnea and occasional palpitations for one month. On evaluation at a tertiary care centre, found to have dilated LA/LV with severe MR and was referred to our institute at 38 weeks gestation. Subsequently delivered a male baby boy with birth weight of 2.7 kg via normal vaginal delivery. On Postpartum day 2, she had severe chest tightness, dyspnea and orthopnea with hypotension and tachycardia. She was transferred to cardiac ICCU and started on diuretics and ionotropic supports. Chest X-ray (Fig. 1) showed cardiomegaly. ECG showed sinus tachycardia with p mitrale and high voltage QRS complex (Fig. 2).

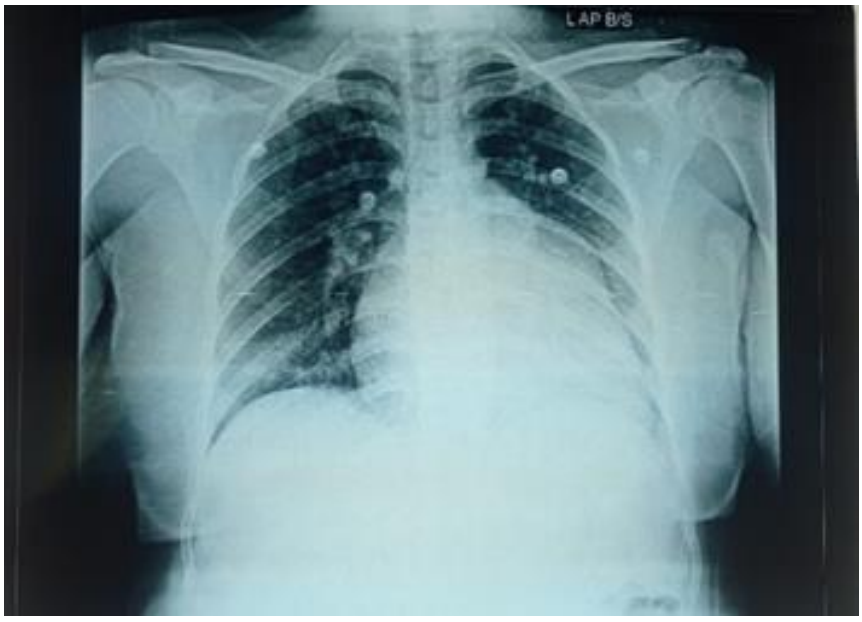

Figure 1: Chest X-ray showing cardiomegaly.

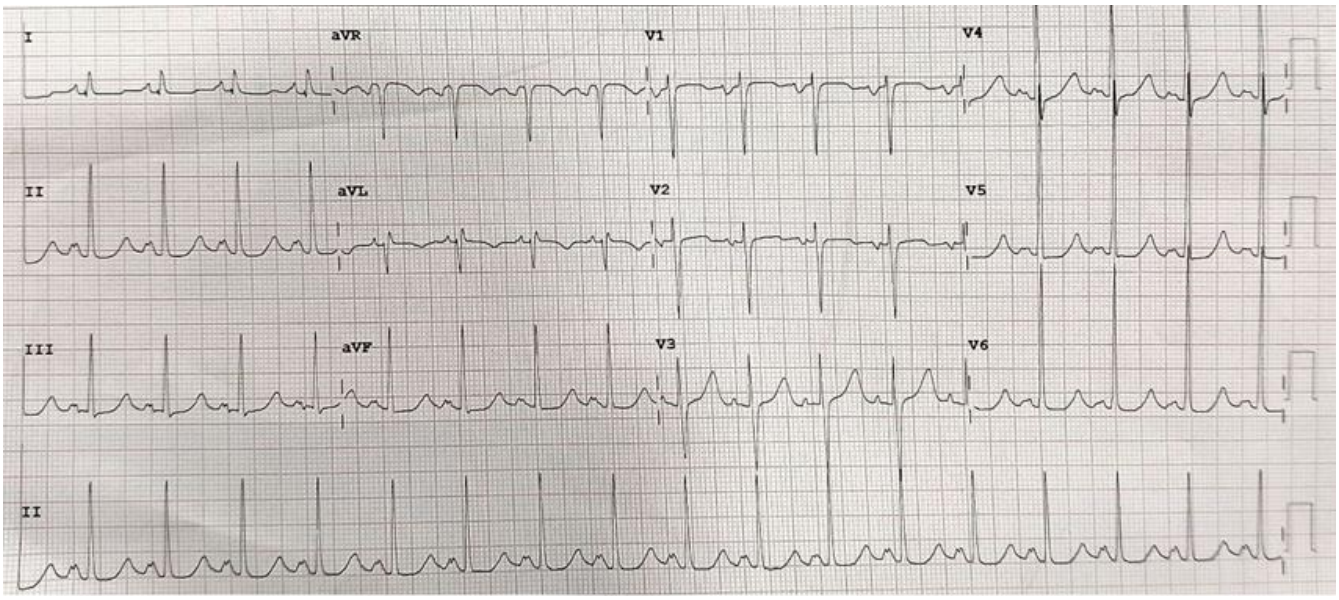

Figure 2: ECG showing Sinus Tachycardia with P mitrale.

Echo (Fig. 3) showed global hypokinesia of LV, severe LV dysfunction with severe MR, thickened mitral leaflets suspecting rheumatic etiology. On further evaluation, TEE showed Dilated LA/LV, severe PAH, severe LV dysfunction with severe MR - thickened anterior mitral leaflet and mildly restricted posterior mitral leaflet. The above findings combined with severe LV dysfunction needed a close therapeutic workup. In order to rule out any underlying cardiomyopathy, patient was further planned for Cardiac MRI. The result showed CRHD- severe MR with non-compaction LV (Fig. 4,5). The etiology for severe MR, couldn't be explained by echocardiography, TEE and Cardiac MRI. Hence it was concluded that severe MR might be due to LV Non-Compaction. Based on above findings, she was not a suitable candidate for MV replacement and hence was discharged on conservative management. Based on all the above investigations and considering the age of the patient, she might be an ideal candidate for cardiac transplantation. The patient improved symptomatically at follow up visit after 2 months, but had significant echocardiographic findings and hence was advised to continue same medical management.

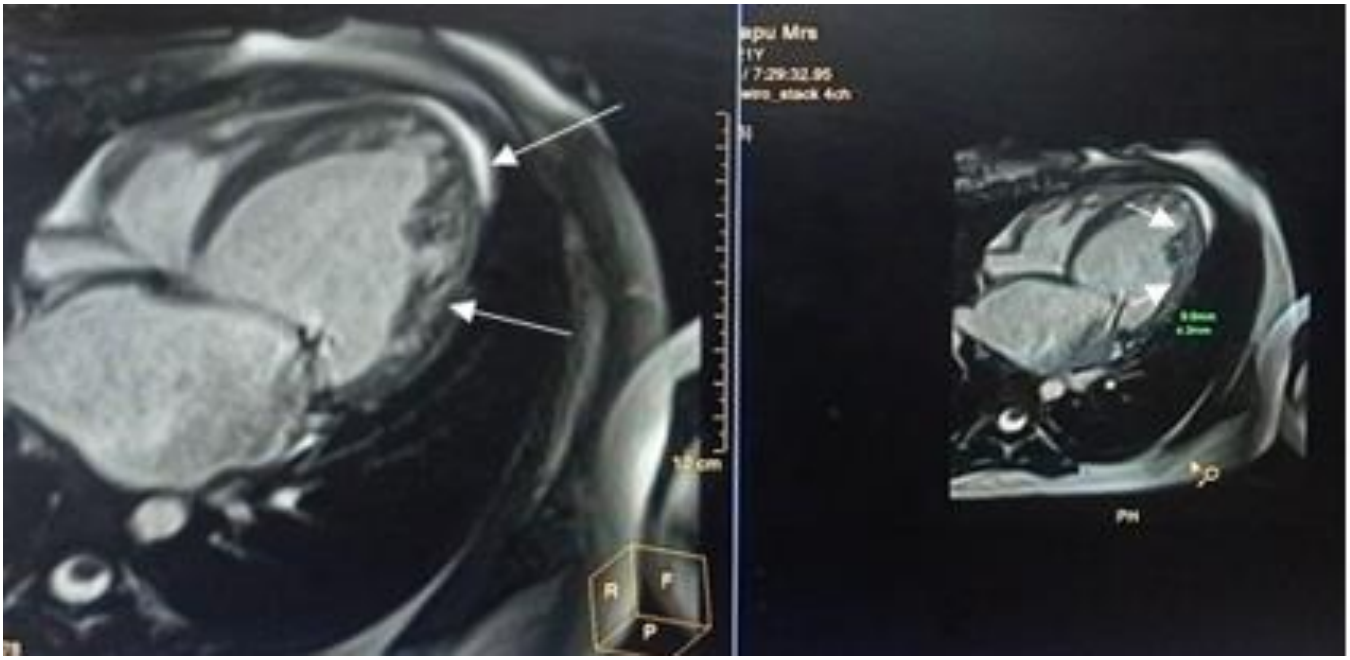

Figure 3: Four chamber view revealing dilated LA/LV with Severe Mitral Regurgitation visualized by colour flow doppler imaging.

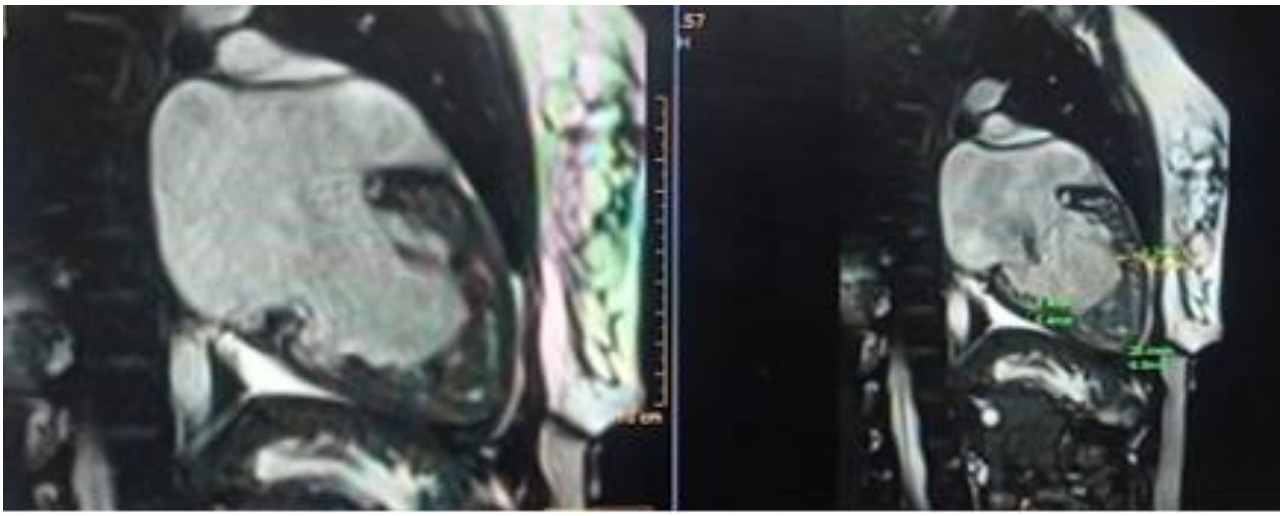

Figure 4: Cardiac MRI- A two chamber bright blood images with Prominent apex and anterior wall.

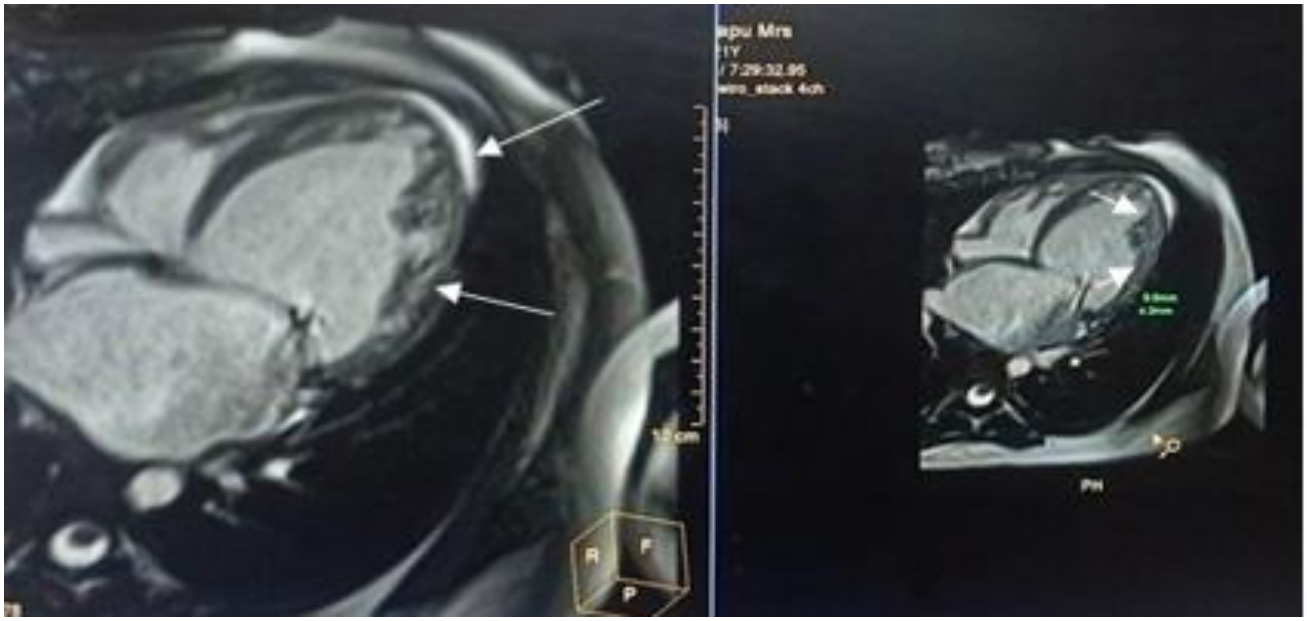

Figure 5: Cardiac MRI- A four chamber view in horizontal long axis revealing septal sparing with Dilated LA/LV and thickened LV lateral wall with prominent trabeculations.

Discussion

Non compaction cardiomyopathy also known as "SPONGY MYOCARDIUM" due to its spongy appearance. Isolated noncompaction cardiomyopathy was first described by Engberding and Bender in 1984. It may be familial or sporadic in nature [6]. The sporadic form occurred in 60-70% of cases. Prominent trabeculations on endocardial surfaces with recesses extending into the left ventricle [7]. Genetic studies show that mutations were more frequent in sarcomere genes whereas non sarcomere gene mutations were identified in rare cases with complex genotypes. The American Heart Association classified it as congenital genetic cardiomyopathy [8].

The myocardium has a trabeculated aspect in embryogenesis, before the development of coronary arteries. The process starts at the base of heart and progresses towards the apex. LVNC develops as a consequence of early cessation of this compaction during embryogenesis [9]. The reason why epicardial layer is compacted and endocardial layer is not because of prominent trabeculae and recesses which communicate with left ventricle. In LVNC there is a mismatch between myocardial mass and number of capillaries leading to hypoperfusion of endocardium, which causes reduced ejection fraction and systolic dysfunction. According to Jenni, et al., diagnostic criteria for LVNC by echocardiography included the ratio of noncompacted to compacted myocardium >2:1. Cardiac MRI can be used in correlation with echocardiography to localize the extent of noncompaction [10]. Petersen, et al.,

criteria elaborated in 2005 have accepted cardiac MRI as diagnostic parameters for the evaluation of LVNC, the presence of two distinct myocardial layers and marked trabeculations with deep intertrabecular recesses within the inner noncompacted layer; a noncompacted/compacted myocardium ratio > 2.3 at end-diastole was considered suggestive.

In patients with LVNC, treatment options vary based on presenting symptoms, ranging from medical management for mild cases to heart transplantation in refractory cases [11]. Our case diagnosed in early postpartum showed worsening of symptoms due to associated MVP with severe MR and severe LV dysfunction. Since the patient was not a suitable candidate for MV repair, she was conservatively managed with beta blockers, ACE inhibitors and diuretics. Monitoring asymptomatic patients is encouraged, whether is the diagnosis is incidental or a familial cause because of possible further complications [12-18].

Conclusion

Although not widely diagnosed, left ventricular Non compaction is a condition that has shown increased prevalence in recent clinical practice mainly due to improvements in echocardiographic techniques and use of cardiac MRI studies. The prognosis is determined by the degree of progression and extent of complications such as heart failure, cardiac arrythmias and systemic embolism. Hence, it is advisable that suspected cases of Non compaction LV should be carefully evaluated by appropriate imaging methods. With this rare presentation, we conclude that Cardiac MRI is helpful to evaluate structural heart diseases.

Conflict of Interests

The authors declare no conflict of interest regarding authorship roles or publication of article.

Acknowledgement

Not applicable

Ethical Statement

Not applicable

Informed Consent Statement

Informed consent was obtained from the subject involved in the study.

Author's Contributions

All the authors have equal contribution and all the authors have read and agreed to the published version of the manuscript.

Financial Disclosure

No funding was not involved in the manuscript writing, editing, approval or decision to publish.

Consent for Publication

Informed consent was obtained from the patient for publication of this case report and is stated in the manuscript.

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