

Isolated Left Ventricle Non-Compaction—Rare Form of Cardiomyopathy—Case Report

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Abstract: Left ventricular Non-compaction (LVNC) is a rare congenital cardiomyopathy, characterized by a thin, compacted epicardial layer, an extensive non-compacted endocardial layer, prominent trabeculation and deep recesses, that communicates with the left ventricular cavity, but not with the coronary circulation^(1,2). This is probably due to an arrest of compaction, during intrauterine life. Clinical manifestations are highly variable, ranging from no symptoms to disabling congestive heart failure, life threatening arrhythmias and systemic thromboembolism.

Keywords: Left ventricle Non-Compaction, non-compaction cardiomyopathy, trabeculation, LVNC

1. Introduction

Left ventricle Non-compaction(LVNC), or 'spongy myocardium' is a rare, morphologically distinct primary genetic cardiomyopathy characterized by prominent ventricular trabeculations and deep inter-trabecular recesses, that is probably due to an arrest in endo-myocardial morphogenesis, leading to persistence of fetal myocardium in postnatal life^(1,2). Non-compaction may present as an isolated cardiomyopathy or may be associated with complex congenital heart lesions. Literature reviews revealed that the prevalence of the disease is between 0.014 and 1.3% in the general population. This condition can potentially lead to chronic heart failure, life threatening ventricular arrhythmias and systemic embolic events. Accurate diagnosis and prompt treatment in such cases may prove lifesaving.

In this article, we report an emblematic case of LVNC, diagnostic modalities, and recent treatment guidelines.

2. Case Report

A 56-year-old nonsmoker, presented with complaints of shortness of breath (NYHA II) for 2 months. He had similar episode 2 weeks prior and was managed conservatively. The symptoms was gradually progressive. He had no history of chest pain, palpitation or syncope. He had no past history COPD, hypertension, or any thromboembolic events in any

other system. He was recently diagnosed to have T2 diabetes mellitus, well controlled with antidiabetic medication.

General examination of the patient revealed a conscious, oriented and afebrile patient with HR - 72beats/min (regular), BP- 110/70mmHg, RR-18/min and SpO2- 95% (room air). Systemic examination revealed S3 gallop, 3/6 pan-systolic murmur at apex, crackles in the bilateral lower lung fields and mild bilateral pedal edema extending up to the knees.

Chest x-ray revealed bilateral oligemic lung fields. Routine lab investigations including cardiac biomarkers were within normal limits, except for elevated NTproBNP - 963.2. ECG showed sinus rhythm with left bundle branch block and ventricular ectopics. 2D Echo (Fig: 1 - 4), revealed global hypokinesia of left ventricle (LVEF-25%), dilated LA/LV, sinuses and trabeculae at apex, features suggestive of probable non-compaction of left ventricle. Confirmation was done by MRI (Fig: 5-7), that revealed two distinct (myocardial) layers having thin compacted Epicardial layer and much thicker non-compacted Endocardial layer. 24 hour-Holter monitoring was done in view of frequent ectopics that showed few couplets, bigeminy, trigeminy and one short run of non-sustained ventricular tachycardia. Diagnostic angiography revealed 30-40% mid LAD stenosis and 30-40% mid RCA stenosis.

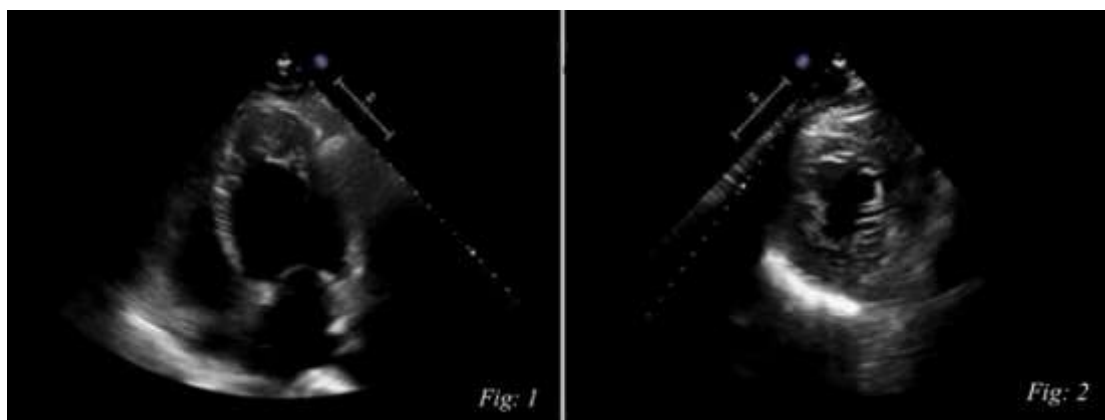


Figure 1: In apical 4-chamber view: left ventricular non-compaction seen as prominent trabeculation in apical segments, **Figure 2:** Parasternal short axis view: trabeculae and recesses are seen in apical segments)

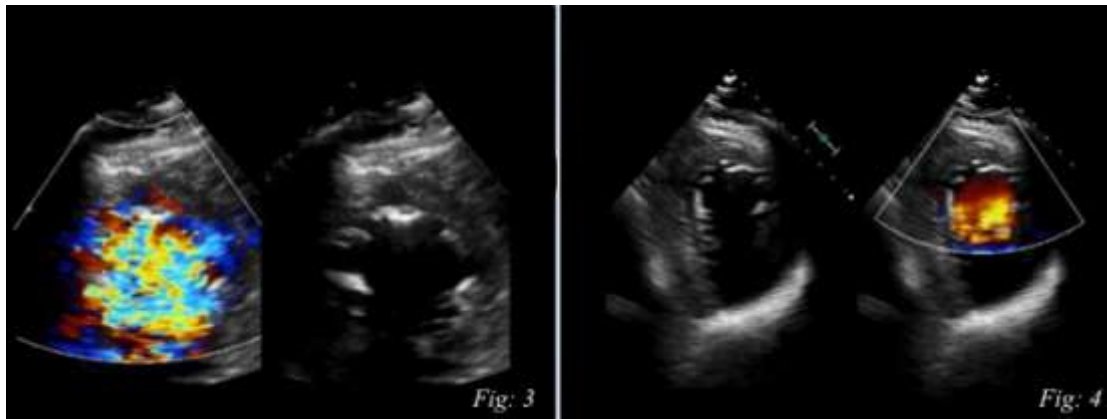


Figure 3 & Figure 4: Color Doppler flow highlights perfusion of inter-trabecular recesses from LV cavity)

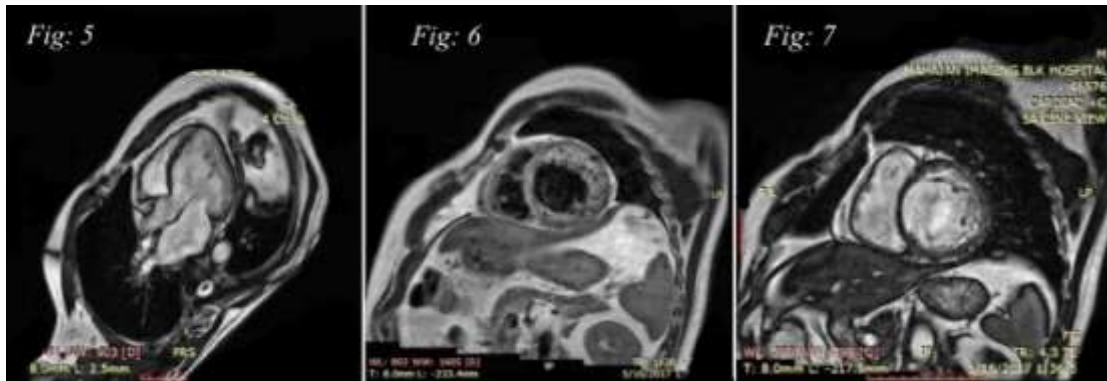


Figure 5: CMRI in long axis view showing Non-compaction in apical segments, **Figure 6, Figure 7:** CMRI in short axis view showing non-compacted to compacted myocardial ratio of 3.6. The thickness of compacted and non-compacted myocardium obtained at end-diastole at mid left ventricular level in short axis cine images. The non-compacted to compacted myocardial ratio is $18.1 : 5 = 3.6$ (Ratio > 2.3 at the end-diastolic is diagnostic for left ventricular non-compaction).

He was started on diuretics (Furosemide), Beta-blockers, ACE-inhibitors and anticoagulation therapy. Single chamber AICD implantation was done for primary prevention of sudden cardiac death or life-threatening arrhythmias as per ACC, AHA and ESC guidelines.

3. Discussion

American Heart Association (AHA) classifies Left Ventricle Non-compaction (LVNC) as a primary genetic cardiomyopathy,^(3,4) while European Society of Cardiology (ESC) describes (LVNC) as an unclassified cardiomyopathy. It is characterized by the presence of an extensive non-compacted myocardial layer lining the cavity of the left ventricle⁽⁵⁾. Echocardiography is the most widely used imaging modality for the diagnosis of LVNC, though the apical region is poorly visualized by echocardiography and can lead to underestimation of the degree of the left ventricular non-compaction⁽⁶⁾. Poor acoustic windows could mislead even experienced echocardiographers the diagnosis of this disease, resulting in an erroneous label such as dilated or hypertrophic cardiomyopathy⁽⁷⁾. Cardiovascular magnetic resonance (CMR) has become the method of choice to confirm or rule out LVNC and may outperform echocardiography in defining the morphology and extension of myocardial non-compaction,^(5,8) that was demonstrated in a study by Yousef et al.⁽⁹⁾ CMR can also give valuable diagnostic and prognostic information about the disease by depicting fibrosis on delayed contrast-enhanced images.⁽⁶⁾

Both isolated forms of LVNC or forms associated with another congenital anomaly⁽¹⁵⁾, cardiac or non-cardiac, have been described. Commonly associated anomalies include ventricular septal defect, coarctation of aorta, transposition of great vessels, and atrial septal defect. The precise pathophysiologic mechanism of the association of LVNC and other congenital heart diseases (including PAVD) has not been elucidated, and more studies are needed in this area.

Patients at the time of diagnosis may be asymptomatic or may present with complications that include heart failure, arrhythmias, and thromboembolic events. Rhythm disturbances include atrial fibrillation, Wolff- Parkinson-White syndrome (WPW syndrome), and ventricular tachycardia and hence, the need for electrocardiograms to diagnose and monitor the occurrence of these potentially lethal conditions.^(5,10) Thromboembolic events that occurs, are due to atrial fibrillation or thrombus formation within the intra-trabecular recesses in the non-compacted myocardium.⁷

Oechslin et al⁽¹¹⁾ reported in a group of 34 adults, with left ventricular non-compaction, presence of higher final diastolic diameter of left ventricle, low ejection fraction, persistent or permanent atrial fibrillation and bundle branch block were related with high risk and poor prognosis, calling to consider the possibility of implantation of an automated cardiac defibrillator and evaluation for transplant. The mortality rate was similar in left ventricular

non-compactation as with patients with non-ischemic dilated cardiomyopathy (3-year survival of 85% vs 83 %).⁽¹²⁾ Diagnosis calls for the study of family members, and genetic counseling. Due to the high prevalence of neuromuscular disorders reported in patients with left ventricular non-compactation, neurological and musculoskeletal evaluations are also recommended.^(13,14)

The classical triad of heart failure, ventricular arrhythmias and systemic embolic events, which were initially reported in patients with advanced disease, are less frequently observed in recent studies.

4. Conclusion

Left ventricular non-compactation is a rare cardiomyopathy, but that should always be considered as a possible diagnosis because of its potential complications. Echocardiography is the standard tool for diagnosis, and CMR is confirmatory, especially when the apex is difficult to visualize. Nevertheless, there are still many questions regarding this disease and further studies are required to elucidate management strategies and thereby reducing the morbidity and mortality rates in such cases.

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