Noncompaction Cardiomyopathy: A Catastrophic Cause of Heart Failure

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INTRODUCTION

- Left Ventricular Noncompaction Cardiomyopathy (LVNC) is a relatively rare congenital disorder characterized by prominent trabeculations and intertrabecular recesses that communicate with the ventricular cavity.[1,2]
- The clinical presentation can be variable. Patients can be asymptomatic or have ventricular arrhythmias, thromboembolism, heart failure, and even present with sudden cardiac death.[3]

CASE PRESENTATION

- Patient is a 40 year old male with a history of Non Ischemic Cardiomyopathy (NICM), Heart failure with reduced ejection fraction, Left Ventricular (LV) thrombus, Atrial fibrillation, and Stroke who presented with an acute heart failure exacerbation.
- He was taking carvedilol and warfarin. Also, he was wearing a LifeVest for primary prevention of sudden cardiac death in the setting of severely reduced EF.
- Transthoracic echocardiogram on this admission demonstrated a **LVEF of 10-15%**, severe mitral & tricuspid regurgitation, and a stable LV apical thrombus compared to prior studies. **Prominent trabeculae in the left ventricle were noted as seen in *Figure 1*.**
- Cardiac MRI was performed for further evaluation. It showed evidence of **borderline LV noncompaction with a noncompacted:compacted myocardium ratio of 2.1:2.2 as shown in *Figure 2*.**
- Spironolactone, Sacubitril/Valsartan, and Empagliflozin were started for optimization of his guideline-directed medical therapy
- He was referred for genetic counseling to evaluate for noncompaction cardiomyopathy gene variants in both himself and his first degree relatives
- If he fails medical therapy, evaluation for left ventricular assist device (LVAD) and consideration for heart transplant could be pursued.

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Figures and Imaging

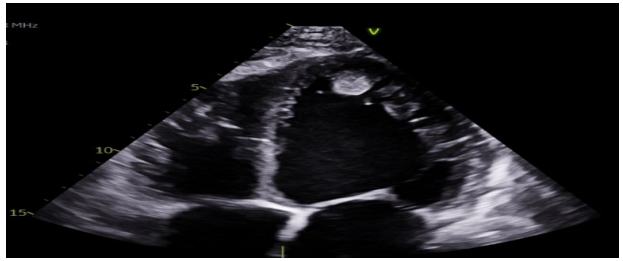

Figure 1. NICM with trabeculae involving the midventricular & apical segments with an apical thrombus

Diagnostic Echo Criteria based on the Jenni Criteria:

(i) Bilayered myocardium with multiple, prominent trabeculations in end-systole
(ii) NonCompacted/Compacted ratio > 2:1
(iii) Communication with the intertrabecular space demonstrated with color doppler
(iv) Absence of coexisting cardiac abnormalities.

Diagnostic MRI Criteria

Pathological noncompaction: NC/C >2.3 in end-diastole
Specificity and NPV: 99%

Table 1.[4]

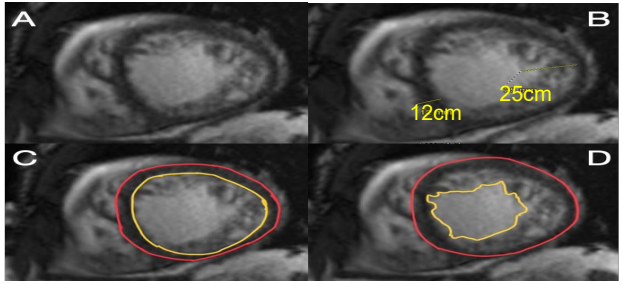

FIGURE 2: Cardiac MRI **A** short axis view left ventricle; **B** maximal left ventricular noncompaction:compaction ratio of 2.1-2.2:1; **C** epicardial and endocardial contours; **D** epicardial and noncompacted contours.

DISCUSSION

- LVNC is a complex congenital myocardial disorder that occurs due to failure of left ventricular compaction during embryogenesis.
- Etiology: Point mutation in the beta-myosin heavy chain gene.
- Diagnostic criteria for LVNC is shown in *Table 1*.
- *The trabeculations associated with LVNC promote thrombus formation, which can lead to strokes & other thromboembolic phenomena.*
- LVNC is an independent risk factor for malignant arrhythmias and LV dysfunction.[5]
- Management involves treating symptomatic heart failure, LV dysfunction, and arrhythmias. Prophylactic anticoagulation may be warranted in the absence of a LV thrombus, especially in patients with LV dysfunction.[6]

CONCLUSION

- LVNC is an inherited cardiomyopathy which is likely underdiagnosed. **Due to its autosomal dominant inheritance, screening of first degree relatives is recommended**.[7]
- Through our poster & our case, we aim to increase the awareness of this disease and its complications: LV thrombus and stroke.

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