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Left Ventricular Noncompaction Syndrome: A Rare Congenital Cardiomyopathy

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Left ventricular noncompaction syndrome: a rare congenital cardiomyopathy

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Abstract

Isolated left ventricular noncompaction cardiomyopathy (LVNC) is a rare congenital condition occurring due to arrest of myocardial compaction in the first trimester, resulting in a thin layer of compacted epicardium and thick hypertrabeculated myocardium containing deep recesses. This article presents a 44-year-old female with progressive dyspnea, orthopnea, paroxysmal nocturnal dyspnea, and pleuritic chest pain. Examination revealed elevated jugular venous distention, lower extremity edema, and bibasilar crackles on lung auscultation, while the electrocardiogram (EKG) exhibited left bundle branch block. Two-dimensional echocardiography (2D-Echo) showed a dilated left ventricle (LV) with ejection fraction (EF) of 25% and severe diffuse hypokinesia. Cardiac magnetic resonance imaging (MRI) confirmed the diagnosis of LVNC. Thereafter, she developed atrial fibrillation with rapid ventricular rate. Conservative treatment was initiated with Tikosyn, Xarelto, Lasix, Toprol XL, Lisinopril, statin and life vest. Eventually, a cardiac resynchronization therapy defibrillator (CRT-D) was implanted to prevent sudden cardiac death and reduce heart failure complications.

Keywords

noncompaction, cardiomyopathy, congenital, heart failure

Introduction

Isolated Left Ventricular Noncompaction Cardiomyopathy (LVNC) is a relatively rare congenital condition, which may be familial or sporadic, resulting from arrest of the normal compaction process of the myocardium during the first trimester of fetal development. This subsequently produces a thin layer of compacted epicardium and a thick hypertrabeculated myocardium containing deep recesses. It is commonly misdiagnosed and it has high morbidity and mortality if not diagnosed early and managed aggressively.

Case Presentation

A 44-year-old white female presented to the emergency room with a two-week history of progressive shortness of breath, orthopnea, paroxysmal nocturnal dyspnea, bilateral leg swelling, and chest pain with deep inspiration. Her past medical history included deep vein thrombosis; she denied a history of coronary artery disease, hypertension, diabetes mellitus, and hyperlipidemia. She was a current and longtime smoker at the time of admission. In family history, her mother had atrial fibrillation, congestive heart failure and coronary artery disease. The physical examination showed stable vital signs, an elevated jugular venous distention and edematous lower extremities. Lung auscultation revealed bilateral basal crackles, while the electrocardiogram (EKG) exhibited left bundle branch block.

The labs were significant for an elevated B-type natriuretic peptide and D-Dimer, whereas the complete blood count and basic metabolic profile were within normal limits. Chest radiograph showed pulmonary congestion consistent with congestive heart failure. Chest computed tomography (CT) was done, which ruled out pulmonary embolism, and venous Doppler revealed evidence of chronic deep venous thrombosis in the left leg.

She was initially treated conservatively with an intravenous diuretic, oral beta blocker, ACE-inhibitor and statin. During the second day after hospitalization, she developed atrial fibrillation with rapid ventricular rate and telemetry showed multiple episodes of non-sustained ventricular

tachycardia. Two-dimensional echocardiography (2D-Echo) exhibited a dilated left atrium, severely trabeculated left ventricle (LV) with ejection fraction (EF) of 25% and severe diffuse wall hypokinesia (Figure 1). Cardiac magnetic resonance imaging (MRI) confirmed the diagnosis of LVNC with a ratio of 3.2/1.¹ (Figure 2).

Figure 1: Echocardiography of left ventricle showing thick myocardium with deep recesses.

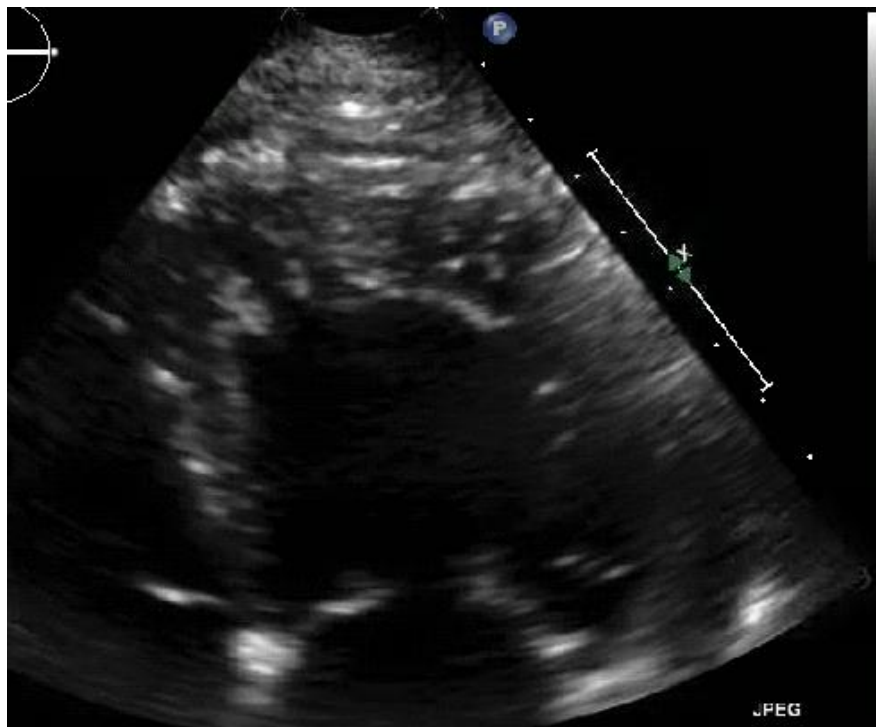


Figure 2: Cardiac MRI showing thick myocardium of left ventricle.



The electrophysiologist recommended aggressive rhythm control with Dofetilide due to ventricular arrhythmias, after which the patient was discharged home with a life vest. Eventually, cardiac resynchronization therapy defibrillator (CRT-D) implantation was performed to prevent sudden cardiac death and to reduce morbidity and mortality related to heart failure.

Discussion

The American Heart Association classifies LVNC as a primary genetic cardiomyopathy, which has been reported in both children and adults. The prevalence in adults is 0.01-0.27%.^{2,3}

During the first trimester of pregnancy, the heart consists of a trabeculated meshwork, which has deep recesses within the myocardium, and blood is supplied through the intertrabecular space. At 5-8 weeks of gestation, the ventricular muscles undergo compaction of the trabeculated meshwork and form a solid myocardium.⁴ In LVNC, the normal compaction process of the myocardium arrests, resulting in a thin layer of compacted epicardium and a thick hypertrabeculated myocardium with deep recesses.

The most common presentation includes symptoms of congestive heart failure. It could present as systolic dysfunction due to decreased subendocardial perfusion or diastolic dysfunction due to abnormal relaxation and restrictive filling due to the trabeculae. It could also present as global LV dysfunction due to mechanical incoordination between the thick trabeculated myocardium and thin epicardium. Additional sequelae include atrial fibrillation in 25%, ventricular arrhythmias in 47%, and sudden cardiac death in 40% of LVNC patients.⁵

LVNC is most commonly diagnosed with 2D echocardiography. There are currently numerous non-standardized criteria to diagnose LVNC by echocardiography. In our case report, echocardiography exhibited a severely trabeculated apex and low EF of 25%. The diagnosis was then confirmed by cardiac MRI which showed LV myocardial trabeculation with a noncompacted to compacted ratio of 3.2/1. A ratio of 2.3 during diastole has a sensitivity of 86% and specificity of 99%.¹

There is currently no specific treatment for LVNC. Management involves standard medical therapy for the treatment of congestive heart failure with low EF. Symptomatic ventricular arrhythmias and low EF are treated with anti-arrhythmic drugs and an implantable cardiac defibrillator (ICD) due to high rates of sudden cardiac death in these patients. Atrial fibrillation and chronic anticoagulation therapy are recommended in patients with EF <40% and a history of thromboembolism, as they are at high risk for thromboembolic episodes due to thrombus formation within the deep intratrabecular recesses. Finally, heart transplantation is reserved for patients with refractory symptomatic heart failure provided there are no contraindications for the procedure.⁶ Fricker et al described indications for heart transplantation in children: children < 6 months who are hospitalized on inotropic support and have little chance of survival or adolescents, if LVEF is <25% or echocardiographic left ventricular internal diameter is > 7cm.⁷ Since LVNC is associated with high rates of morbidity and mortality, an aggressive workup and management are required in patients who are suspected to have this condition.

In the pediatric population, LVNC is usually associated with other congenital abnormalities. Thus, isolated LVNC is difficult to diagnose in early life, with a prevalence of 0.14%.⁸ Specifically, amongst children with primary cardiomyopathy, it was found that LVNC was present in 9.2% of patients.⁹ Even in children, symptoms of heart failure are the most common

form of presentation; additional symptoms include chest pain, syncope, failure to thrive, cyanosis, an abnormal cardiac exam, abnormal EKG and echocardiogram. They may have transient recovery of ventricular function during childhood, then present later in adulthood as late presentations due to the deterioration of LV function.¹⁰ Wolff-Parkinson-White syndrome and ventricular tachycardia are common arrhythmias in the pediatric population.

In summary, LVNC is a genetically heterogeneous congenital disorder assumed to be due to arrest in early embryogenesis. The clinical presentation ranges from asymptomatic to heart failure, atrial and ventricular arrhythmias, thromboembolism, and sudden cardiac death. It is often a misdiagnosed cause of cardiorespiratory failure. Since the disease is associated with high morbidity and mortality, increased awareness is vital for early diagnosis, which significantly influences the decisions of long-term management, prognosis, and the need to screen living relatives.

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