Case Report

Noncompaction Cardiomyopathy: Case Presentation with Cardiac Magnetic Resonance Imaging Findings and Literature Review

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ABSTRACT

Left ventricular noncompaction cardiomyopathy is a very rare condition, yet believed to be often overlooked. It is thought to be caused by the developmental arrest in embryogenesis and characterized by an increase in the noncompacted, trabeculated myocardium adjacent to compacted myocardium in the left ventricular. The clinical presentations of this type of cardiomyopathy are of variable severity. Echocardiography used to be the diagnostic modality, but recent reports suggest that cardiac magnetic resonance imaging has higher sensitivity and specificity by showing a ratio of the noncompacted myocardium to compacted myocardium of >2.3.

Key words: Cardiac magnetic resonance, cardiomyopathy, left ventricular noncompaction, magnetic resonance, noncompaction

INTRODUCTION

Left ventricular noncompaction cardiomyopathy (LVNC) is a very rare cardiomyopathy characterized by an increase in the noncompacted, trabeculated myocardium adjacent to compacted myocardium in the left ventricular (LV).[1-3] Though current research suggests a developmental arrest in embryogenesis as the underlying pathology,[2,4] the etiology of LVNC are not fully understood. The patients will often present with a spectrum of disease severity ranging from no symptoms to cardiac arrhythmias, cardiac failure, thromboembolism or even sudden cardiac death.[4-6]

Historically, LVNC has been diagnosed by echocardiography when the ratio of noncompacted to compacted myocardium is >2. Echocardiography may not visualize the apical region optimally, leading to underestimation of the degree of LV noncompaction.[3] However, cardiac magnetic resonance imaging (CMRI) provides a comprehensive depiction of cardiac morphology in any imaging plane. Recent CMRI reports suggest a ratio of the noncompacted myocardium to compacted myocardium of >2.3 yield the highest sensitivity (86%) and specificity (99%) in diagnosis.[7]

We present a case of LVNC in an adult patient with its clinical and imaging findings.

CASE REPORT

A 24-year-old female patient was initially referred to our vascular department for investigation of bilateral lymphedema. Her echocardiogram showed mildly impaired LV with evidence of segmental wall motion abnormality and hypertrabeculation in the apex suggestive of noncompaction. Further questioning revealed a history of sudden death in two of her brothers. One of the deaths was related to an unknown cardiomyopathy. The history of the patient started at birth when she had difficulty swallowing and several delayed milestones such as walking and talking at the age of 5. Due to socioeconomic issues, the parents did not seek medical assistance. As the patient matured, there was an additional history of palpitations, which were occasionally associated with syncopal attacks.

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The patient had generalized, but progressive weakness throughout the years. Her weakness increased with activity and decreased with a rest. The weakness was mainly proximal, worse in the upper limbs. Due to constant fatigability, she dropped out of school.

Her vital signs were normal on physical examination, she demonstrated normal heart sounds. Lower limbs bilateral nonpitting edema up to the knees, proximal bilateral upper and lower limbs weakness with intact sensation, waddling gait, and easy fatigability with few steps were noted.

Computed tomography coronary angiogram was performed to exclude coronary artery disease. Results showed segmental wall motion abnormality and revealed normal coronary arteries, LV dilatation, and lateral wall and apical noncompaction with the noncompacted myocardium to compacted myocardium ratio in the range of 2.5–3.5 [Figure 1]. CMRI was done for further evaluation of LV function and volume. Her CMRI showed mildly dilated LV with mild global hypokinesis and a marked decrease in longitudinal shortening. End-diastolic volume was 120 mL, end-systolic volume was 61 mL and the ejection fraction was 50%. In addition, multiple areas of noncompacted myocardium, particularly in the apex and most of the lateral wall extending from apical lateral wall to the mid-to-basal segment were seen. The ratio of maximum thickness of the noncompacted myocardium to compacted myocardium was more than 2.5 in multiple areas [Figure 2]. Mildly dilated left atrium and mild mitral regurgitation were seen. Right ventricle (RV) has a prominent trabeculation, but normal systolic function.

Further investigation showed elevated creatine kinase and electromyography findings consistent with myopathy. Left deltoid biopsy showed advanced dystrophic changes of undetermined type. Brain magnetic resonance imaging (MRI) was negative.

Thus, the clinical impression was proximal myopathy mostly mitochondrial or congenital, congenital lymphedema, LV noncompaction with LV ejection fraction of 40%, and normal RV function. As her LV impairment is mild, the decision was made to keep her on beta-blockers and on angiotensin-converting enzyme inhibitor in the form of metoprolol and lisinopril, respectively. On subsequent follow-up visits, she reported a history of interval improvement apart of occasional palpitations with no history of shortness of breath, syncope or dizziness. She has been advised to do regular physiotherapy and to wear compressive stockings for her lymphedema.

DISCUSSION

Noncompaction cardiomyopathy used to be called “spongy myocardium” due to its spongy appearance.[4] In 1932, Bellet and Gouley described the first case of noncompaction in an autopsy of a newborn infant with aortic atresia and coronary–ventricular fistula.[8] LVNC without other cardiac abnormalities (isolated noncompaction cardiomyopathy) was first described in 1984. Historically, the prevalence of LVNC has been underestimated due to the lack of knowledge about this rare condition and its similarity to other diseases of the myocardium and endocardium. However, diagnosis of LVNC has increased over the last 25 years. A study of pediatric patients with primary cardiomyopathy showed that 9% of the patients had LVNC.[9]

The median age at diagnosis of isolated noncompaction cardiomyopathy in the initial case series of isolated noncompaction was 7 years ranging from 11 months to 22 years.[2] The prevalence of LVNC in the adult population ranges between 0.01% and
0.3% of all adult patients referred for echocardiography studies.\textsuperscript{[10–12]} These numbers are mostly extracted from the referred patient for abnormal echocardiographic findings or congestive heart failure (HF). Therefore, because of this potential selection bias, the true prevalence is not clear. Male patients are predominately affected accounting for 63%, 70%, and 74% of cases in three different reported series.\textsuperscript{[2,5,13]} 

In 2001, Jenni et al. evaluated seven patients with LV noncompaction.\textsuperscript{[19]} Histologic analysis demonstrated ischemic lesions in the thickened endocardium and thickened trabeculae with accompanying fibrosis. Clinical manifestations are highly variable, ranging from no symptoms to disabling congestive HF, arrhythmias, and systemic thromboembolism.

According to a French registry, the diagnosis of LVNC diagnosis was confirmed in 105 cases through the use of echocardiogram performed in a laboratory between 2004 and 2006.\textsuperscript{[15]} In that study, LVNC was first detected in 12 patients with rhythm disorders, 45 patients with HF symptoms, and eight patients through familial screening. During the follow-up study, patients suffered several complications including HF occurring in 33 of the patients, ventricular arrhythmia in seven, embolic events in nine, and nine of the patients received heart transplantation while death occurred in 12 of the patients.

A Swiss registry recorded a total of 34 cases over 15 years among a patient population who underwent echocardiogram.\textsuperscript{[13]} In this study, major complications included HF (53%), ventricular tachycardia (41%), thromboembolic events (24%), and death (35%). Six of the reported 12 deaths were sudden and four due to end-stage HF in four and the other two cases were due to unrelated causes. Four patients underwent heart transplantation and four patients had automated cardiovert/defibrillators.

The presented case had a family history of sudden death in two brothers, with one of them presumed to have an unknown type cardiomyopathy. Sporadic and familial forms of noncompaction have been described. In the original report of isolated noncompaction ventricular cardiomyopathy that predominantly affected children, half of the patients had a familial recurrence.\textsuperscript{[2]} However, a larger reported adult population with isolated noncompaction ventricular cardiomyopathy showed only 18% familial recurrence.\textsuperscript{[13]} Lower percentage compared with the earlier report might be attributed to incomplete screening of siblings in Oechslin et al. study group. This patient has a progressive myopathy, which raised the question of other association with her left ventricular noncompaction cardiomyopathy. About 82% of LV noncompaction cases were found to be associated with neuromuscular disorder\textsuperscript{[1]} including Becker muscular dystrophy, metabolic myopathy, myotonic dystrophy, Barth syndrome, and other rare genetic disorders.\textsuperscript{[9]}

The diagnosis of LVNC is often made by echocardiography. The ratio of noncompacted myocardium to compacted myocardium at the end of systole is $>2:1$ is the most often used echocardiographic criteria that were proposed by Jenni et al. The criteria also include presence of segmental thickening of the myocardial wall of the LV with two layers: Prominent trabeculations and deep recesses with a thin epicardial layer and a thick endocardial layer. The absence of coexisting cardiac abnormalities is required to fulfill the criteria. LV diastolic dysfunction, reduced global LV systolic function, abnormal structure of papillary muscles, and LV thrombi are nonspecific findings that can be seen on echocardiography.\textsuperscript{[14]}

Cardiac magnetic resonance (CMR) can be used in correlation with an echocardiogram to localize and quantify the extent of noncompaction. CMR offers a detailed view of the cardiac morphology of the noncompacted myocardial layer in the LV in any image plane including the apical and lateral segments-segments, which are not well visualized by echocardiogram.\textsuperscript{[7]} Recent reports suggest that echocardiography diagnostic criteria are strict and MRI enhances the detection of more subtle forms of noncompaction.\textsuperscript{[16–18]} Further, CMR identifies a higher rate of two-layered structures in segments such as the anterior, inferior, anterolateral, and inferolateral segments compared with echocardiogram.\textsuperscript{[7]} However, contraindication after implanted devices, cost and availability of CMR are considered barriers on the way of implementing CMR.

According to the American Heart Association, 1.5 Tesla is the minimum acceptable magnet strength to perform cardiac imaging in order to be able to visualize both short and long axis views in approximately 17 segments. Acquiring three diastolic long axis views best identifies the NC/C ratio of the most prominent myocardial trabeculations segment. The NC/C ratio in CMR is measured at the end of diastole and should be more than 2.3, more than echocardiography, which is 2.1. Stagnant blood flow, within the myocardial trabeculae may be detracted as a high signal in CMR black blood imaging, supporting the diagnosis LVNC.\textsuperscript{[19]}

The efficacy of CMR was evaluated in a report of seven patients with LVNC in whom other features supported the diagnosis; the results were compared with 170 healthy volunteers, athletes, or patients with dilated cardiomyopathy or hypertrophic cardiomyopathy, aortic stenosis, and hypertensive heart disease. The most distinguishing feature was a ratio of noncompacted to compacted myocardium during diastole (sensitivity 86% and specificity 99%).\textsuperscript{[20]} Direct imaging of myocardial fibrosis is possible with the use of an inversion recovery prepared T1-weighted gradient-echo sequence
and the extracellular fluid tracer gadopentetate dimeglumine.[20] This technique has been termed “delayed hyperenhancement” and shows nonviable tissue as hyperenhanced or bright.

Unfortunately, no treatment exists for LVNC. Nevertheless, the early and precise diagnosis is mandatory to rule out other underlying diagnoses and to allow a timely start of standard HF and anticoagulation therapy, thus preventing further complications.

CONCLUSIONS

The incidence of reported noncompaction ventricular cardiomyopathy has been increasing in the literature. The clinical presentations are of variable severity and the exact prevalence is unclear. Thus, it tends to be overlooked. CMRI delayed hyperenhancement sequences could demonstrate delayed trabecular hyperenhancement. Some trabeculae show delayed hyperenhancement despite having a normal compacted myocardial ratio, suggesting that LV noncompaction may be a more diffuse disease process than previously suspected. The use of delayed hyperenhancement sequences improves the correlation between CMRI and the parameters of the clinical stage of the disease. CMRI follow-up examinations would be helpful to assess a potential change of noncompacted or compacted mass in a chronological sequence. In conclusion, CMR can distinguish LVNC from other cardiomyopathies and normal hearts with high sensitivity and specificity.

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