CASE REPORT

Cardiomyopathy Following Biventricular Noncompaction

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ABSTRACT

Myocardial noncompaction (NC) is usually seen isolated; however, sometimes other congenital heart abnormalities may accompany the myocardial noncompaction. Left ventricular noncompaction (LVNC) is a genetic anomaly where the ventricular wall is replaced by thick ventricular trabeculations with deep intertrabecular recesses held together by a thin compacted layer. The most common site of involvement is the left ventricle, with right ventricular involvement being reported in a few cases. Isolated right ventricular noncompaction (RVNC) is rare yet life-threatening if left untreated. Genetic testing may identify possible mutation of gene. Early diagnosis of NC is very important for disease management. The management of associated other cardiac pathologies simultaneously will help improve the symptoms and prognosis in patients with noncompaction. Here we report a case of 60-year-old male patient presenting with heart failure due to cardiomyopathy with biventricular noncompaction. The case is being presented as an academic interest.

Keywords: *Biventricular noncompaction, cardiomyopathy, biventricular cardiac failure, congenital anomaly of heart*

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INTRODUCTION

Myocardial noncompaction (NC) is usually seen isolated; however, sometimes other congenital heart abnormalities may accompany the myocardial noncompaction.[1] Left ventricular noncompaction (LVNC) is a genetic anomaly where the ventricular wall is replaced by thick ventricular trabeculations with deep intertrabecular recesses held together by a thin compacted layer.[2] The most common site of involvement is the left ventricle, with right ventricular involvement being reported in a few cases.[3] Isolated right ventricular noncompaction (RVNC) is rare yet may become life-threatening if left untreated. During embryogenesis, portions of myocardium fail to compact correctly. This leaves areas of the wall of the ventricles with a loosely compacted network that does not pump blood effectively.[1] Clinical manifestations are highly variable, ranging from no symptoms to disabling congestive heart failure, arrhythmias, and systemic thromboemboli.[4,5] Echocardiography has been the diagnostic procedure of choice, but the correct diagnosis is often missed or delayed because of lack of knowledge about this uncommon disease and its similarity to other diseases of the myocardium and endocardium.[5] Here, we present the case of a 60-year-old male patient who was admitted with heart failure and later diagnosed with cardiomyopathy with biventricular

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noncompaction. The case is being presented as an academic interest. Written informed consent was obtained from the patient for publication of this case report and accompanying images.

CASE SUMMARY

A 60-year-old male patient was admitted into the Department of Cardiology, Mymensingh Medical College Hospital, Bangladesh, with the complaints of shortness of breathing for two months along with generalized swelling and palpitations. He did not complain of any chest pain or syncope. Physical examination releveled bilateral pitting oedema with pulmonary crackles and a 2/6 systolic murmur heard loudest at the left lower parasternal border during auscultation. The laboratory results were within normal limits. Electrocardiogram (ECG) showed ST-T changes and LBBB pattern with occasional premature ventricular complexes (Fig. 1). Chest x-ray P/A view showed gross cardiomegaly (Fig. 2). Cardiac evaluation with echocardiogram (Fig.3-7) showed depressed left ventricular systolic function (ejection fraction 30-35%) and impaired right ventricular function (TAPSE-10mm), biatrial enlargement, and increased left ventricular (LV) and right ventricular (RV) wall thickness. The left and right ventricular wall showed marked trabeculation within the inner layer of myocardium consistent with LV and RV noncompaction cardiomyopathy. Contrast study showed blood flow through the trabeculated noncompact inner layer to the outer compact layer. Moderate mitral and tricuspid regurgitation and mild pulmonary hypertension were noted (PASP 35 mmHg). After admission, he was treated with oral and injectable form of diuretics and ACE inhibitor. The patient was symptomatically improved within three days. Then he was discharged with further advice to do a cardiac MRI and genetic study (e.g., *TTN* mutation) in a specialized centre.

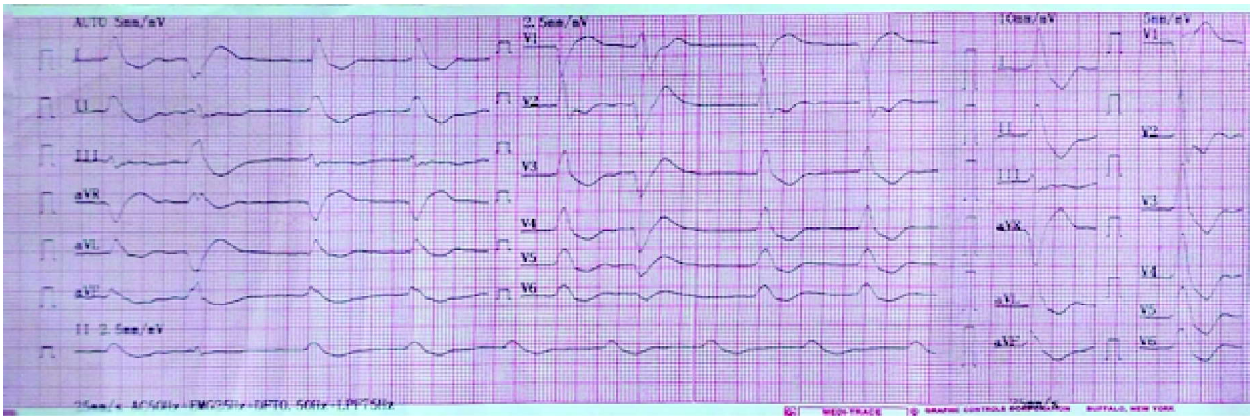

Fig. 1: *ECG on admission, showing LBBB pattern, and multiple premature ventricular complexes.*

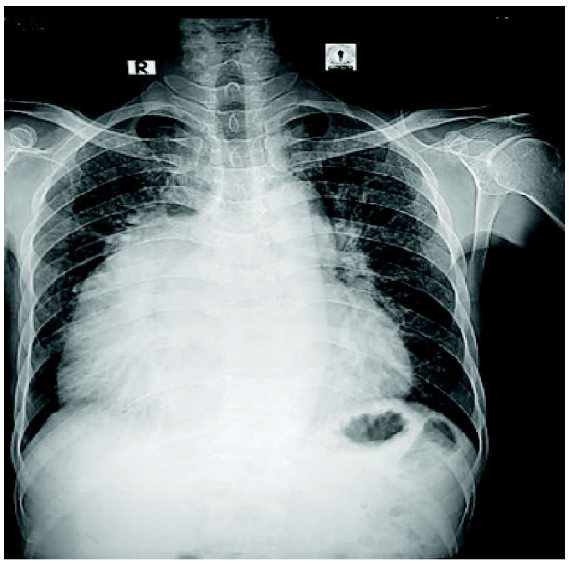

Fig. 2: *Chest x-ray P/A view shows gross cardiomegaly.*

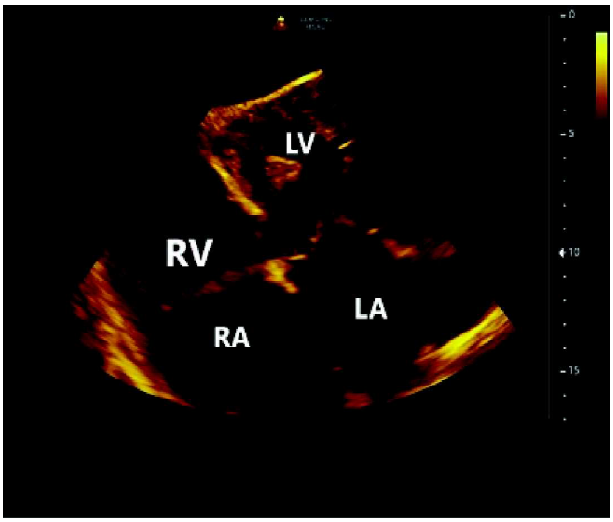

Fig. 3: *Echocardiography showing noncompacted LV and RV.*

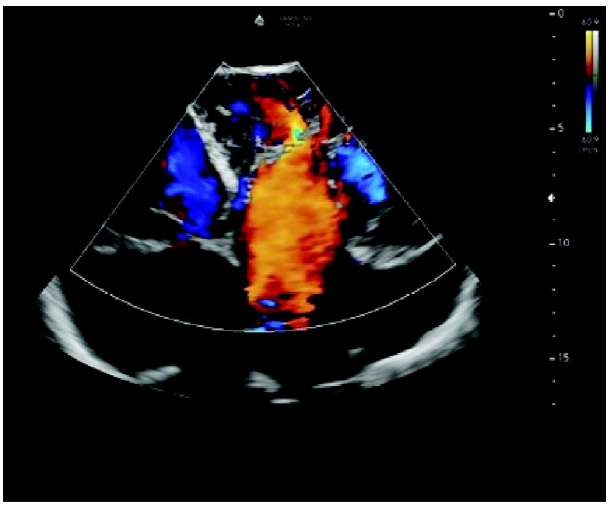

Fig. 4: *Echocardiography showing blood flow into intertrabecular recesses.*

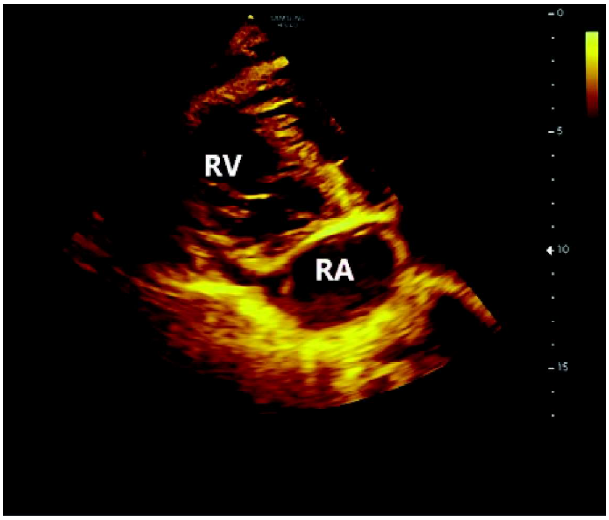

Fig. 5: *Echocardiography showing noncompacted RV.*

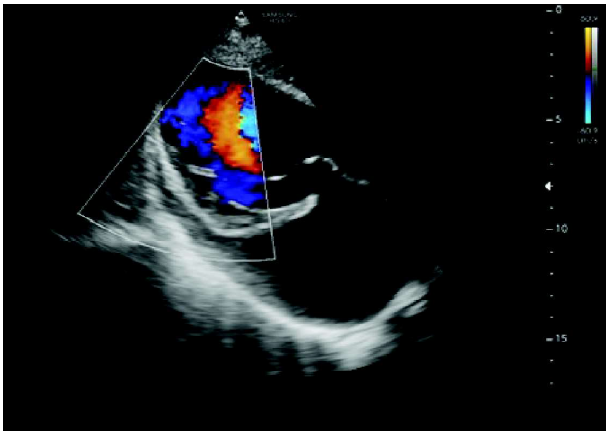

Fig. 6: *Echocardiography showing blood flow into intertrabecular recesses of RV.*

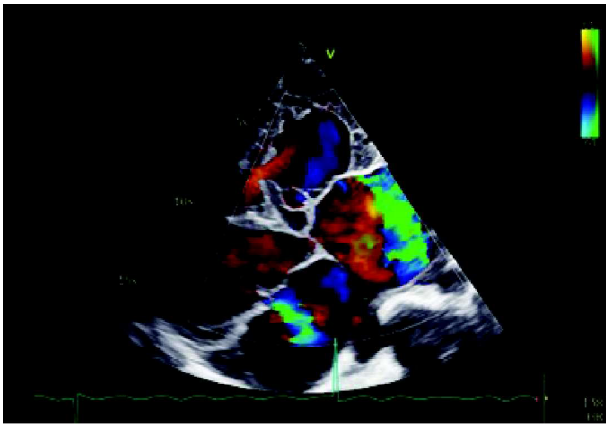

Fig. 7: *Echocardiography showing MR and TR.*

DISCUSSION

Noncompaction of the ventricular myocardium is a cardiomyopathy caused by the arrest of normal embryogenesis of the ventricles. It is classified in isolated noncompaction of the ventricles (most frequently of the left one) and in ventricular noncompaction associated with other congenital anomalies of the endocardium and myocardium.[6] It is characterized by an altered myocardial wall with prominent trabeculae and deep intertrabecular recesses. This results in a thickened bilayer of compacted and noncompacted myocardium following the arrest of the normal process of endomyocardial morphogenesis.[7] Both familial and sporadic forms of non-compaction have been described. Although genes responsible for the sporadic forms have not yet been identified, genes responsible for some familial cases of VNC have been described and have been linked to a mutation in the G4.5 gene of Xq28 chromosome region.[4] Males appear to be affected more often than females, with males accounting for 56%-82% of cases.[5,8] VNC can be present at any age.[8,9] Three major clinical manifestations of VNC are congestive cardiac failure (CCF), arrhythmias (atrial arrhythmias, ventricular tachycardia and sudden cardiac arrest) and thromboembolic events; however, findings vary among patients, ranging from asymptomatic left ventricular dysfunction to severe disabling CCF.[5-9]

There are three proposed diagnostic criteria that are most utilized in the literature. Chin et al.[10] are given credit for their first attempt to define specific criteria for the diagnosis of LVNC. The evaluation includes left ventricular (LV) free-wall thickness at end-diastole, prominent trabeculations, and a progressive

decrease in the ratio of myocardial thickness from the epicardial surface to the trough (*X*) and the epicardial surface to the peak (*Y*) of the trabeculations in the PSAX and apical views.[10] Stöllberger et al.[11] refined the definition as >3 trabeculations protruding from the LV wall apical to the papillary muscles, perfused intertrabecular spaces, and a two-layered myocardium with the noncompacted layer usually thicker than the compacted myocardium in end-systole. However, many recent studies followed criteria set by Jenni et al.[12] to evaluate the presence of LVNC. These criteria include a bilayered myocardium, a noncompacted to compacted ratio >2:1, communication with the intertrabecular space demonstrated by Doppler, absence of coexisting cardiac abnormalities, and presence of multiple prominent trabeculations in end-systole.[12] The echocardiographic criteria proposed by Jenni et al.[12] are also frequently followed. Those include:

(1) the absence of coexisting cardiac anomalies;

(2) the presence of a two-layered structure of the Left Ventricle wall, with the end-systolic ratio of the non-compacted to compacted myocardial layer greater than two, measured in parasternal short axis view;

(3) finding this structure predominantly in the apical and mid-ventricular areas; and

(4) blood flow directly from the ventricular cavity into deep intertrabecular recesses as assessed by Doppler echocardiography or intravenous contrast.[12]

CONCLUSION

Noncompaction is a rare congenital cardiomyopathy which has a poor prognosis. The diagnosis of NC can be made with echocardiogram once the clinician has elicited a thorough family history coupled with a high degree of clinical suspicion. Early diagnosis of NC is very important for disease management. The management of associated other cardiac pathologies simultaneously will help improve the symptoms and prognosis in patients with noncompaction.

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