Noncompaction of the ventricular myocardium with tetralogy of Fallot

Fallot tetralojisi ile "noncompacted" ventriküler miyokard

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Introduction

Noncompaction of the ventricular myocardium (NVM) is a rare congenital unclassified cardiomyopathy. It is caused by a defect in cardiac morphogenesis resulting in an arrest of compaction of loose interwoven meshwork of myocardial fibers during intrauterine life (1). It can result with severe systolic dysfunction as well as hypertrophy of the involved walls of the ventricles (2). It is diagnosed both in children and adults. Currently, the incidence of this rare cardiomyopathy is estimated 0.05% in adults (3). The disorder is usually diagnosed by two-dimensional echocardiography. It can be seen in patients with other congenital anomalies or in isolated form in the absence of associated anomalies (4). We describe an illustrative case of NVM, a 22-year-old male with tetralogy of Fallot (TOF) and progressive worsening of heart failure.

Case report

A 22-year-old man was evaluated at the outpatient clinic for dyspnea and weakness on exertion. He had TOF and Blalock-Taussig shunt was done 12 years ago. There was a history of sudden cardiac death (his father and brother) in his family. Mild cyanosis, protodiastolic gallop, apical 4/6 systolic murmur, hepatomegaly and jugular venous pulse were revealed on physical examination. Patient systolic/diastolic blood pressures and heart rate were 110/70 mmHg and 102 beats/min respectively. Sinus tachycardia and nonspecific interventricular conduction delay (QRS duration 140 ms) with a right heart axis were seen on electrocardiography. Cardiomegaly without pulmonary congestion and dextraposition of the aorta were revealed on the telecardiography. Two-dimensional transthoracic echocardiography showed dextraposition of aortic root, large membranous ventricular septal defect resembling a single ventricle (Fig. 1), infundibular pulmonary stenosis (gradient 20/14 mmHg), mild mitral regurgitation and severe tricuspid regurgitation (right ventricular (RV) systolic pressure 108 mmHg) and biventricular global hypokinesia (left ventricular (LV) ejection fraction %25, RV ejection fraction %30). The LV, RV end-diastolic and left atrial diameters were 63, 46 and 47 mm respectively. The left and right ventricles appeared to have concentric hypertrophy with multiple, prominent myocardial trabeculations and deep intertrabecular recesses communicating with the main ventricular cavity (Fig. 2 and 3). The end-systolic ratio of noncompacted to compacted myocardium was greater than 2:1. Accordingly, diagnosis of NVM was established. Left and right-sided heart catheterization was performed (systolic/diastolic pulmonary artery pressure 98/36 mmHg, RV pressure 114 mmHg, right atrial pressure 9 mmHg, pulmonary capillary wedge pressure 22 mmHg and pulmonic flow/systemic flow ratio: 1.1). The patient was taken under medical therapy (including silazapril 5 mg/d, carvedilol 6.25 mg/d, aspirin 150 mg/d, furosemid 20 mg/d and spironolacton 50 mg/d) and he is currently being evaluated for heart and lung transplantation.

Discussion

Myocardial noncompaction is a rare form of cardiomyopathy due to an arrest in endomyocardial embryogenesis (5). Noncompacted ventricular myocardium may be associated with other congenital anomalies, such as obstruction of the right or left ventricular outflow tracts, complex cyanotic congenital heart disease, and coronary artery anomalies (6,7). Pressure overload or myocardial ischemia in these secondary cases of myocardial noncompaction may prevent the normal regression of embryonic myocardial sinusoids. However, currently it is unclear whether the isolated and secondary forms of noncompaction are common or distinct lesions.

In the present case, NVM was associated with TOF and although the outflow tract obstruction was in the RV, both of the ventricles were affected by noncompaction. The patient had a low gradient in the right outflow tract and pulmonary hypertension. This hemodynamic situation is very rare in a TOF and low RV ejection fraction and/or increased pulmonary vascular resis-

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tance may be the reasons.

Both familial and sporadic forms of NVM have been described. Familial recurrence was seen in half of patients (4). Likewise, sudden cardiac death occurrences existed in his family history. In X-linked form of the disease, a locus has been found on Xq28, and mutations have been reported in G4.5 gene (4).

Noncompacted ventricular myocardium is usually detected by echocardiography and confirmed by cardiac magnetic resonance imaging. The characteristic echocardiographic findings consist of multiple, excessively prominent myocardial trabeculations and deep intertrabecular recesses perfused from the ventricular cavity, commonly involving the apical and mid-ventricular segments of the left ventricle (8).

Prominent myocardial trabeculation can be found in healthy hearts as well as in hypertrophic myocardium secondary to cardiomyopathies, valvular heart disease or hypertension. Thus, the differentiation between variants and NVM may often be challenging. An end-systolic ratio of non-compacted to compacted layers of >2 is diagnostic for NVM and allows differentiation of the trabeculations of NVM from that observed with dilated cardiomyopathy or hypertensive cardiomyopathy (9). In addition, evidence of direct blood flow from the ventricular cavity into deep intertrabecular recesses by colour Doppler echocardiography is one of the hallmarks of the diagnosis of NVM. This feature is clearly never been observed in other forms of LV hypertrophy (9).

In our case, echocardiography revealed biventricular dilatation with severely impaired systolic function. Characteristic, multiple, prominent muscular trabeculations were present in the both of the ventricles. The end-systolic ratio of noncompacted to compacted myocardium was greater than 2:1. Deep intertrabecular spaces communicating with the main ventricular cavity were evident on both two-dimensional and colour flow imaging. These findings were pathognomonic for NVM.

The clinical manifestations include congestive heart failure signs, arrhythmias and cardiac embolic events. The most common presentation is congestive heart failure, as in our case. Prognosis is poor and the common causes of death are intractable heart failure and sudden cardiac death (10). The end-stage congestive heart failure is managed with heart transplantation.

In summary, we present a coexistence of NVM and TOF in a 22-year-old male with the typical clinical and echocardiographic features of the disease.

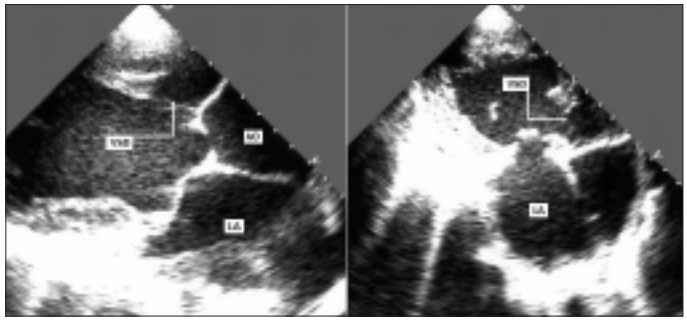

Figure 1. Two-dimensional echocardiography views of large membranous ventricular septal defect

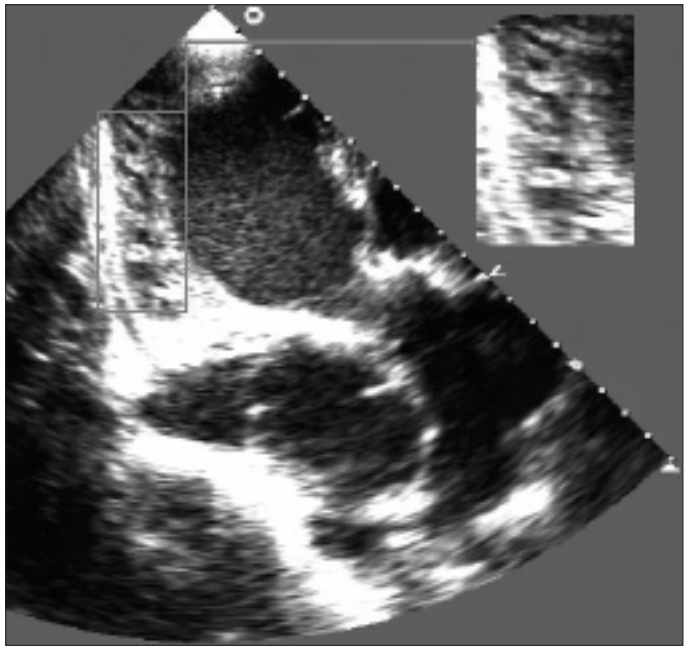

Figure 2. Two-dimensional echocardiography view of multiple prominent myocardial trabeculations

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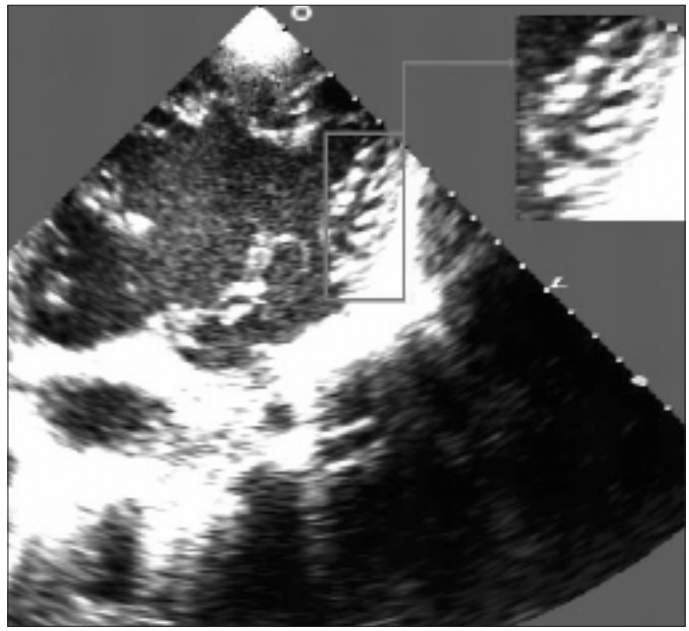

Figure 3. Two-dimensional echocardiography view of deep intertrabecular recesses

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