



Noncompaction of the ventricular myocardium with bicuspid aortic valve

Biküspid aort kapağı ile görülen ventriküler miyokarda “noncompaction”

Yüksel Çavuşoğlu, Recep Aslan, Alparslan Birdane, Demet Özbabalık**, Necmi Ata*

From Departments of Cardiology, Cardiovascular Surgery*, and Neurology**,
Faculty of Medicine, Eskişehir Osmangazi University, Eskişehir, Turkey

Introduction

Noncompaction of the ventricular myocardium (NVM) is a rare congenital disorder thought to be due to an arrest of the physiologic intrauterine compaction process of the ventricular myocardium during embryonic heart development (1). The disease is characterized by multiple prominent trabeculations with deep intertrabecular recesses perfused from the ventricular cavity (2). Currently, diagnosis is based on the findings of transthoracic (TTE) and transesophageal (TEE) echocardiography. Although this rare disorder is defined as a congenital genetic cardiomyopathy and usually is diagnosed in pediatric population (3), it has become more frequently identified in adult patients (4). Noncompaction of the ventricular myocardium may occur as an isolated cardiac lesion, as well as it has been described in association with structural heart defects (1, 5-6). In adults, associated valve anomalies have not been previously emphasized. We described a case of NVM in a 19-year-old male with a moderate aortic regurgitation and bicuspid aortic valve.

Case report

A 19-year-old male with the diagnosis of Sydenham's chorea and rheumatic heart disease was referred to the cardiology department for the surgical assessment of aortic regurgitation. Six months earlier, he began to have purposeless, rapid, nonrepetitive involuntary movements of the extremities. However, there was no clear history of the other symptoms and clinical manifestations of acute rheumatic fever. The patients did not have chest pain, palpitations, syncope episode, or systemic embolic events, but had exercise intolerance. Physical examination revealed systolic murmur at the base of the heart and early diastolic decrescendo murmur along the left sternal border.

Electrocardiography demonstrated sinus rhythm with intraventricular conduction delay. Cardiomegaly was present on chest radiography. Transthoracic echocardiography showed left ventricular (LV) dilatation with normal systolic function. LV end-diastolic and end-systolic diameters were 60 mm and 39 mm, respectively. Left ventricular ejection fraction was normal (63%). The left

and right atriums, and the right ventricle were normal. Transthoracic echocardiography showed mild thickening of the aortic valve cusps. However, mitral valve structure was found to be of normal appearance. A moderate aortic regurgitation and mild mitral regurgitation were detected by color Doppler examination. Doppler-determined peak systolic gradient of the aortic valve was 28 mm Hg. In addition to these echocardiographic findings, prominent muscular trabeculations and deep intertrabecular spaces communicating with the ventricular cavity were observed in the infero-postero-lateral region and apical portion of the LV chamber (Fig. 1 and Fig. 2) (See corresponding video/movie image at www.anakarder.com). Patient underwent TEE for the assessment of aortic valve disease and myocardial noncompaction. It clearly demonstrated the presence of characteristic, multiple, prominent myocardial trabeculations and numerous recesses with penetration into the LV wall, involving especially the infero-postero-lateral region. The right ventricle had normal trabecular pattern. A bicuspid aortic valve with a moderate aortic regurgitation was detected during TEE (Fig. 3). Cardiac catheterization with coronary angiography was performed to determine the intracardiac pressures and the degree of aortic regurgitation. Coronary vessels were normal at coronary angiography and there was no congenital coronary anomaly. Left ventricular end-diastolic pressure was 20 mmHg and peak- to-peak systolic aortic-left ventricular gradient was found to be 15 mmHg. Right heart catheterization showed normal pulmonary artery, right ventricular and right atrial pressures and 1+ mitral regurgitation were observed in left ventriculography. Angiographic assessment of aortic regurgitation demonstrated 2+ regurgitation. Neurological assessment including cerebral magnetic resonance imaging confirmed the diagnosis of Sydenham's chorea. The skeletal muscle enzymes were found to be in normal range. However, underlying etiologic reason of aortic valvular disease was considered as bicuspid aortic valve rather than rheumatic fever because of the normal appearance of mitral valve structure. The patient was treated with angiotensin converting-enzyme inhibitor and valproate sodium. Echocardiographic examinations did not show any finding of noncompacted myocardium in the first-degree relatives of the patient.

Discussion

Noncompaction of the ventricular myocardium has recently been categorized as primary genetic cardiomyopathy in the report of the American Heart Association Scientific Statement on the contemporary definitions and classification of the cardiomyopathies (1). Although NVM has been described as congenital genetic cardiomyopathy with depressed systolic function and LV dilatation, in most adult patients, noncompaction of the ventricular myocardium has been reported to diagnose in asymptomatic period and have had a longer clinical course with gradual depression of systolic function and gradual LV dilatation (5-6). The disease might become clinically overt during childhood and adolescence, and symptoms onset may delay until 7th decade (5-6). In our patient NVM was detected in asymptomatic period of the disease.

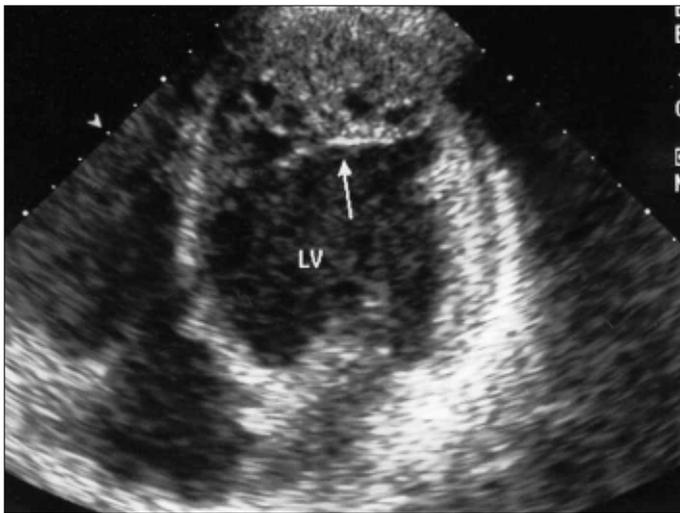


Figure 1. Transthoracic echocardiography shows the multiple, prominent trabeculations and intertrabecular recesses in the apical and in the middle portion of the left ventricle (arrow)

LV- left ventricle

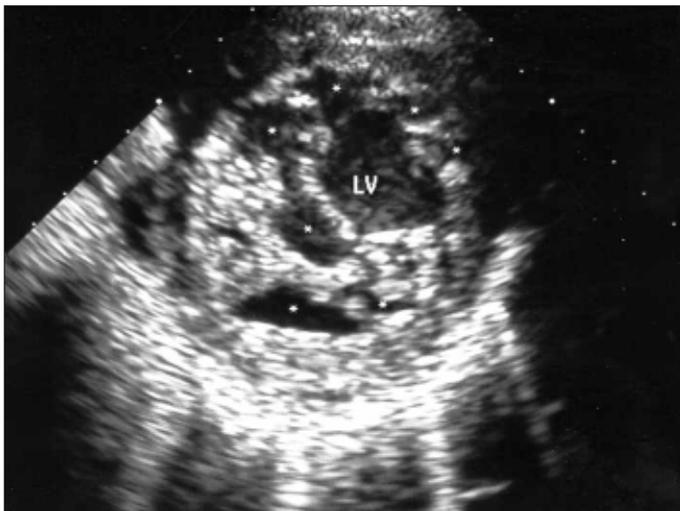


Figure 2. Transthoracic echocardiography. In the short-axis view of the left ventricle, infero-postero-lateral regions are characterized by the presence of recesses and numerous trabeculations

LV- left ventricle

The persistence of the prominent trabecular meshwork of sponge-like myocardium is the constant characteristic findings of ventricular noncompaction. Transthoracic echocardiography is considered as a reliable diagnostic modality for the detection of this characteristic pattern. In patients with non-echogenic picture or poorly defined endocardial borders by TTE, TEE is the alternative method. Furthermore, use of contrast echocardiography may facilitate visualization of the prominent myocardial trabecular recesses and enhance the diagnosis of NVM (7). The 2-layered myocardial wall consisting of a thin compacted epicardial zone and a thickened noncompacted endocardial zone, and the end-systolic thickness ratio between noncompacted and compacted myocardium ≥ 2 , and also intertrabecular recesses perfused from the LV cavity confirmed by the color Doppler technique are used to diagnostic echocardiographic criteria. Noncompacted to compacted layer ratio calculated at the level of the papillary muscles was 2.6 in the patient presented in this report. Magnetic resonance imaging, computed tomography and ventriculography can also be useful imaging modalities (5). However, NVM is often misdiagnosed since it is not widely known. The disease is identified more frequently as an incidental finding during routine echocardiographic evaluations. Therefore, the echocardiographer should be aware of this often unrecognized congenital disorder. In our case, ventricular noncompaction was diagnosed during routine echocardiographic examination performed for the evaluation of aortic valve disease.

Noncompaction of the ventricular myocardium is frequently reported in association with other cardiac and extracardiac disorders. These associated conditions are usually described in pediatric population. In adults, associated cardiac anomalies are rarely reported presumably because these associated anomalies would result in childhood manifestations (6). Neuromuscular disorders are the most frequent noncardiac abnormalities associated with NVM (2). Among the cardiac abnormalities, atrial and ventricular septal defects are the most frequent, reported in five and six patients, respectively, of 223 cases (5%) in a published review (2). The other associated cardiac anomalies include left coronary artery originating from the pulmonary trunk, coronary arterioventricular fistulae, right coronary artery atresia, pulmonary

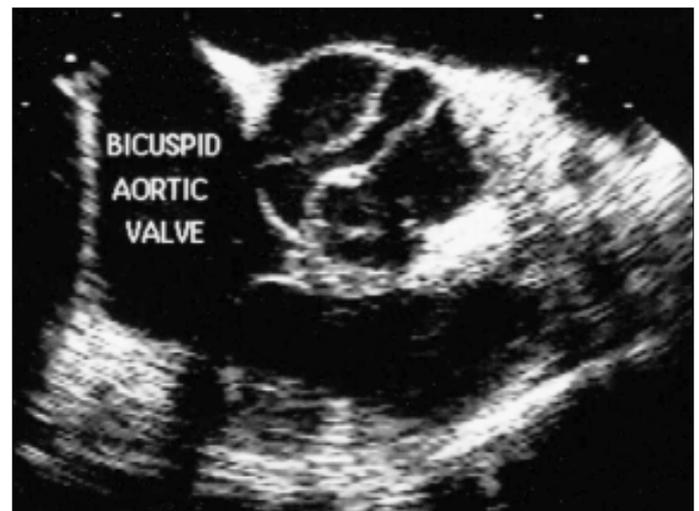


Figure 3. Transesophageal echocardiography. Short-axis view at the level of the aortic valve demonstrates bicuspid aortic valve

artery atresia, subpulmonary stenosis, anomalous pulmonary venous return, patent ductus arteriosus, tetralogy of Fallot, transposition of the great arteries, dextrocardia and aortic atresia (8,9). The concomitant congenital valvular abnormalities described to date consist of mitral atresia, double-orifice mitral valve, mitral valve cleft, congenital mitral valve stenosis, aortic stenosis, absent aortic valve and tricuspid atresia, which all have been primarily reported in infants and children (2,5,8). A case of NVM with bicuspid aortic valve and aortic regurgitation showing poor prognosis had been previously described from our group (10). Recently, a pathological study of 14 cases of infants with NVM and unexpected death has reported a high incidence of valvular anomalies (5/14 cases, 35%) than is currently being appreciated, including bicuspid pulmonary valve, pulmonary stenosis, dysplasia of mitral, pulmonary and tricuspid valves (11). In this autopsy series (11), all the cases representing heart failure have been reported to have associated valve anomalies.

In conclusion, the present case report emphasizes that the concurrent congenital valvular anomalies may be more prevalent in patients with NVM than in general population. Therefore, associated congenital valve anomalies should be investigated in NVM as an associated condition contributing to ventricular dysfunction.

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