Late detection of noncompaction of the myocardium in an adult with complete interventricular septal defect

Tam interventriküler septal defektli erişkin bir hastada geç saptanan süngerimsi miyokart

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Noncompaction of the ventricular myocardium (NVM) is a morphogenetic anomaly that leads to the development of cardiomyopathy. It is often associated with other congenital cardiac malformations. Common clinical presentations of NVM involve heart failure symptoms, ventricular tachyarrhythmias, and thromboembolic events. Although the peculiar echocardiographic picture is characteristic for this entity, it may often be misdiagnosed. In this case report, we describe a 27-year-old man who had been followed-up since childhood, with the diagnosis of interventricular septal defect or enlarged "single ventricle" with a very small rudiment of the apical part of the interventricular septum. On his last echocardiographic examination, NVM was detected with heavy trabeculations and intertrabecular recesses. This case suggests that physicians should be more patient to detect other congenital abnormalities including NVM, which may influence the clinical and prognostic outcome of these patients.

Key words: Cardiomyopathies; echocardiography; heart defects, congenital; heart ventricles; myocardium/pathology.

Ventrikül miyokardında süngerimsi yapı (noncompaction) kardiyomiyopati gelişimine yol açan morfogenetik bir anomalidir. Başka doğumsal anomalilerle birlikteliği sıktır. Süngerimsi miyokardın yaygın klinik bulguları kalp yetersizliği semptomları, ventrikül taşiaritmileri ve tromboembolik olaylardır. Bu durumun ekokardiyografik tablosu oldukça özgün olmasına karşın, atlandığı olgular da olmaktadır. Bu yazıda, çocukluğundan beri interventriküler septal defekt ya da interventriküler septumun apikal kısmının güdük kalmasıyla karakterize büyümüş "tek ventrikül" tanısıyla takip edilmekte olan 27 yaşında bir erkek hasta sunuldu. Hastanın son ekokardiyografik incelemesinde, belirgin trabeküller ve bu trabeküllerin arasında kalan derin girintilerle karakterize süngerimsi miyokart saptandı. Bu olgudan çıkarılması gereken ders, kardiyologların, süngerimsi miyokart da dahil olmak üzere, eşlik eden doğuştan anomalilerin saptanması konusunda daha fazla sabırlı olmaları gerektiğidir; çünkü, bu durumlar hastaların klinik ve prognostik sonuçlarını etkilemektedir.

Anahtar sözcükler: Kardiyomiyopati; ekokardiyografi; kalp defekti, doğuştan; kalp ventrikülü; miyokart/patoloji.

Interventricular septal defect is one of the most common congenital malformations, whereas the prevalence of "single ventricle" is low.[1] Noncompaction of the ventricular myocardium (NVM) is a disorder of endomyocardial morphogenesis characterized by numerous, excessively prominent left ventricular trabeculae and deep intertrabecular recesses, which together create a spongiform appearance.[2] The diagnostic criteria of NVM are: *(i)* >3 prominent trabeculations protruding from the left ventricular wall with deep intertrabecular recesses; *(ii)* direct blood flow from the ventricular cavity into the intertrabecular recesses as visualized by color Doppler; *(iii)* a ratio of ≥2 between noncompacted and compacted subepicardial layers at end-systole.

Noncompaction of the ventricular myocardium is a rare congenital cardiomyopathy, which represents an arrest in intrauterine endomyocardial morphogenesis. It has recently been included in the 2006 classification of cardiomyopathies as a genetic cardiomyopathy.[3] Noncompaction of the myocardium is misdiagnosed or misinterpreted in a very wide range of cases.[4,5]

We present an adult man who had been followed-up with the diagnosis of interventricular septal

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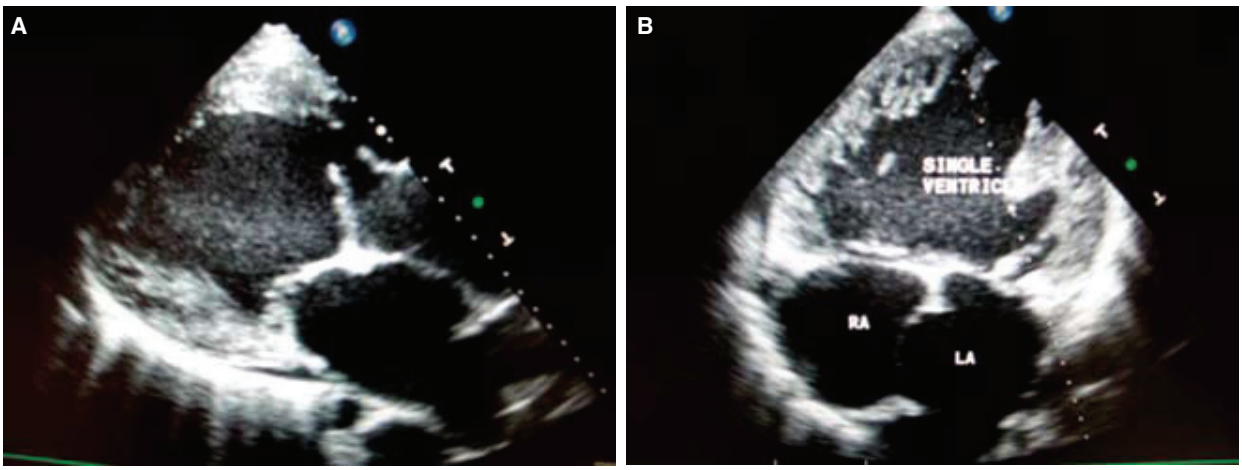

Figure 1. (A) Parasternal long-axis view of two-dimensional echocardiography showing the interventricular septal defect. **(B)** Apical four-chamber view of two-dimensional echocardiography showing the interventricular septal defect and deep intertrabecular recesses.

defect or "single ventricle" since childhood, and whose defect was eventually found to be associated with NVM.

CASE REPORT

A 27-year-old man was admitted to our clinic with complaints of breathlessness at rest, chest pain, palpitations, fatigue, and general asthenia. He was diagnosed, when he was a child, as having interventricular septal defect or "single ventricle", with a very small rudiment of the apical part of the interventricular septum. He had been hospitalized and treated for heart failure in the Department of Cardiology three times during the past three years. He did not have a family history of heart or other hereditary diseases. Physical examination showed a cyanotic face, irregular rhythm, severe systolic murmur at the apex, and rales at the basal lung fields. On admission, the patient's resting electrocardiogram showed atrial fibrillation and right axis deviation, with a ventricular rate of less than 100 beats/min. Blood pressure was 100/70 mmHg. The chest x-ray showed an enlarged heart in all directions and pulmonary vascular congestion. Laboratory analyses showed mild leukocytosis, increased transaminases and urea, decreased level of serum albumins, and decreased oxygen saturation (SaO_2 72%).

There were moderate enlargement of the liver and small amount of ascites on abdominal ultrasound examination, and bilateral pleural effusion on chest x-ray examination.

Two-dimensional transthoracic echocardiography showed a rudiment of the interventricular septum measuring less than one-fourth of its normal presentation, which had been described several times by echocardiographers as "single ventricle", with the dimension of 86 mm in the long-axis parasternal view (Fig. 1a). We estimated the global systolic function of the "single ventricle" as reduced. The left and right atriums were also enlarged; measuring 58 mm and 53 mm, respectively, in the transversal axis. The most important finding in the last echocardiographic examination was typical noncompaction of the "single ventricle" on both sides (Fig 1a, b), with heavy trabeculations and intertrabecular recesses, according to the criteria of Jenni et al.[6] Color-Doppler echocardiography showed mild mitral regurgitation, moderate aortic regurgitation, and severe tricuspid regurgitation with pulmonary artery hypertension (systolic pulmonary artery pressure 95 mmHg) (Fig 2).

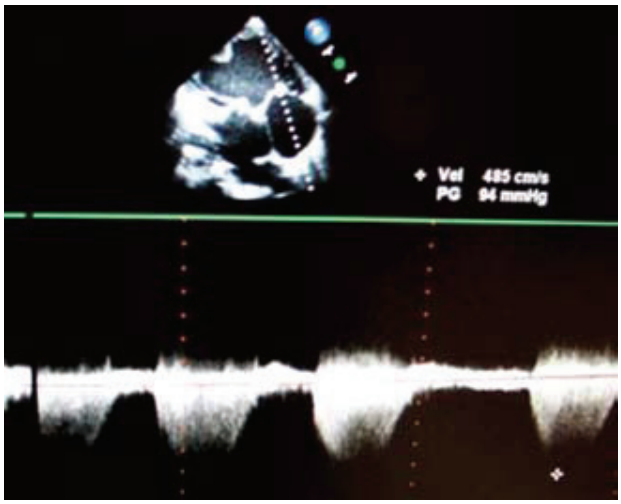

Figure 2. (A) Pulmonary hypertension measured by continuous wave Doppler echocardiography was estimated as >100 mmHg.

The patient was treated with oxygen therapy, beta-blocker, angiotensin-converting enzyme inhibitor, diuretic, digitalis, anticoagulation therapy, and antithrombotics. After two weeks of treatment, he was clinically stable, without breathlessness at rest and with SaO_2 of 86%. After discharge, he was referred for further treatment and eventually heart transplantation in an advanced cardiology center.

DISCUSSION

Ventricular noncompaction is often associated with other congenital heart malformations.[7] Noncompacted myocardium can be considered an inherited congenital malformation since the genes responsible for its development have been identified on chromosome 11p15[8] or as mutations of the gene 4.5 of chromosome Xq28, where other cardiomyopathies have been reported.[9]

Rarely, NVM may occur without associated heart anomalies and is called isolated NVM, a condition that has been described in infants and children, and very rarely in adults.[10]

Noncompaction of the ventricular myocardium uniformly affects the left ventricle, with or without concomitant right ventricular dysfunction and clinical heart failure,[11] ventricular arrhythmias, and embolic events.[12] In our case, the diagnosis of ventricular noncompaction was made based on the characteristic echocardiographic appearance of a two-layered myocardial wall consisting of a thin compacted epicardial and a thick noncompacted endocardial layer with numerous, prominent trabeculations, and deep intertrabecular recesses communicating with the left ventricular cavity.[6,13] The patient had been followed-up since childhood for a complete interventricular septum defect and enlarged "single" ventricle, with a rudiment of the interventricular septum having less than one-fourth of the apical portion. On the last echocardiographic examination, it was diagnosed to be NVM that had not been detected earlier. In a previously published paper by Hughes et al.,[14] among 69 children with a functionally single left ventricle, 15 had the features of noncompaction.

This case suggests that echocardiographers should be more aware of NVM in patients with congenital heart disease. This may influence the clinical and prognostic outcome of these patients.

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