



Asymmetric septal hypertrophy with perimembranous septal defect and obstructive right ventricular outflow tract in a patient with hypertrophic cardiomyopathy

Perimembranöz septal defektli asimetrik septal hipertrofi ile sağ ventriküler çıkış yolu darlığı olan hipertrofik kardiyomiyopati bir hasta

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Introduction

The previous echocardiographic studies have estimated the prevalence of hypertrophic cardiomyopathy (HCM) in the general population to be 0.2% (1). Asymmetric hypertrophy of the left ventricle, predominantly of the septum and anterior wall of the left ventricle, is present in 70 % of patients with HCM. However, right ventricular outflow obstruction with the left ventricular outflow tract intact is a very rare pathology (2). In addition, certain congenital cardiac malformations have been reported in patients with asymmetric septal hypertrophy (ASH) (3). To our knowledge, there are no studies and case reports, which show association between obstructive right ventricular hypertrophy and ASH with perimembranous ventricular septal defect (VSD). We present an ASH case with perimembranous septal defect and obstructive right ventricular hypertrophy.

Case Report

A 27- year-old man presented by himself at our out-patient clinic with dyspnea and exercise intolerance, despite β -blocker therapy. However, no syncope or pre-syncope was present. The patient had no history of rheumatic valve disease, congestive heart failure and hypertension. He was on β -blocker therapy. Physical examination revealed a regular pulse rate of 85 beats/min and a blood pressure of 110/80 mmHg. On his cardiac auscultation, we heard 2-3/6 grade systolic murmur in mesocardiac area, 2/6 grade systolic murmur in pulmonary area, and other physical examination findings were normal. The chest X-ray revealed cardiomegaly and marked pulmonary conus. The electrocardiogram showed sinus rhythm, biventricular hypertrophy, P pulmonale and ST-T changes in precordial and lateral leads.

Transthoracic echocardiography (TTE) revealed small left ventricular size (diastolic diameter: 3.7 cm, systolic diameter: 2.0 cm),

mild biatrial dilatation, perimembranous septal aneurysm with small ventricular septal defect (Fig. 1), asymmetric septal hypertrophy (2.8 cm) (Fig. 2-left), and right ventricular hypertrophy (1.52 cm). No gradient in left ventricular outflow tract (LVOT) blood flow or sings of mitral systolic anterior motion were detected. A systolic turbulent flow with 77 mmHg systolic pressure gradient was detected in the right ventricular outflow tract (RVOT). In addition, mild mitral regurgitation and moderate tricuspid regurgitation were present. Other echocardiographic findings were normal.

In order to confirm these findings, cardiac catheterization was performed by femoral access and revealed similar findings to TTE. No gradient was detected between LVOT and aorta by catheterization. Left ventriculography revealed small left to right shunt. Shunt ratio (Qp/Qs) was 1.07 by the result of oxygen saturation of heart and great vessels. In right cardiac catheterization, there was a significant gradient in RVOT. The catheter findings; the pressures and O₂ saturations of middle right atrium, right ventricle, main pulmonary artery and left ventricle were 10/5/7 mmHg-70%, 105/0/10 mmHg-74%, 25/10/15 mmHg-73% and 130/0/15 mmHg-99%, respectively, and dynamic pressure gradient pattern was seen during pull-back of pressure catheter (peak outflow

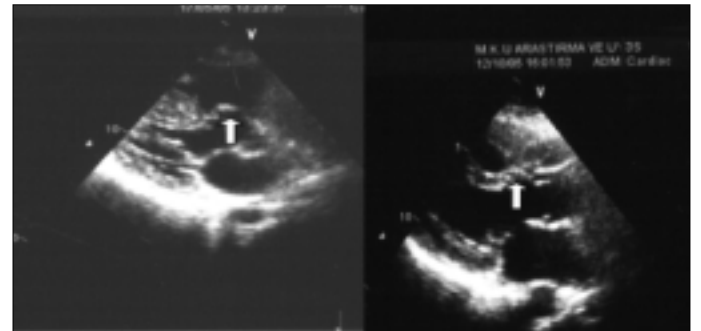


Figure 1. Perimembranous septal aneurysm in parasternal long-axis view before and after operation

gradient: 80 mmHg). Patent ductus arteriosus, coarctation of aorta and other cardiac abnormalities were not detected. The patient was admitted to the hospital for cardiac surgery. Septal myectomy and closure of the perimembranous defect were performed. One month after surgery, transthoracic echocardiography revealed an intact perimembranous septum (Fig. 1), normal interventricular septal thickening (1.0 cm) (Fig. 2b and Fig. 3) and decreased peak RVOT gradient (from 77 mmHg to 20 mmHg). During a follow-up period of 6 months, the patient was asymptomatic.

Discussion

Asymmetric septal hypertrophy is characterized by a disproportionately thickened ventricular septum that contains numerous hypertrophied, bizarrely-shaped and disorganized cardiac muscle cells (3). Although HCM is classically considered a disease of the left ventricle, right ventricular (RV) abnormalities have also been reported. Right ventricular obstruction was present in approximately 15% of patients who have HCM (4).

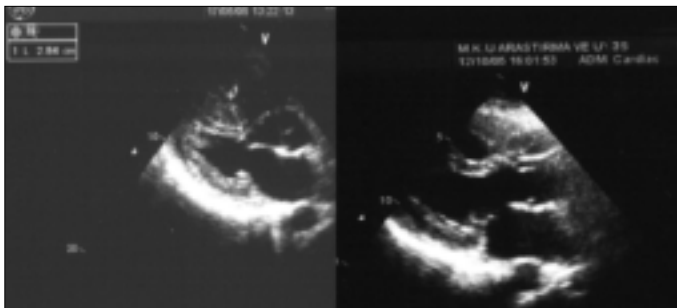


Figure 2. Interventricular septal thickening (2.8 cm vs 1.0 cm) in parasternal long-axis view before and after operation

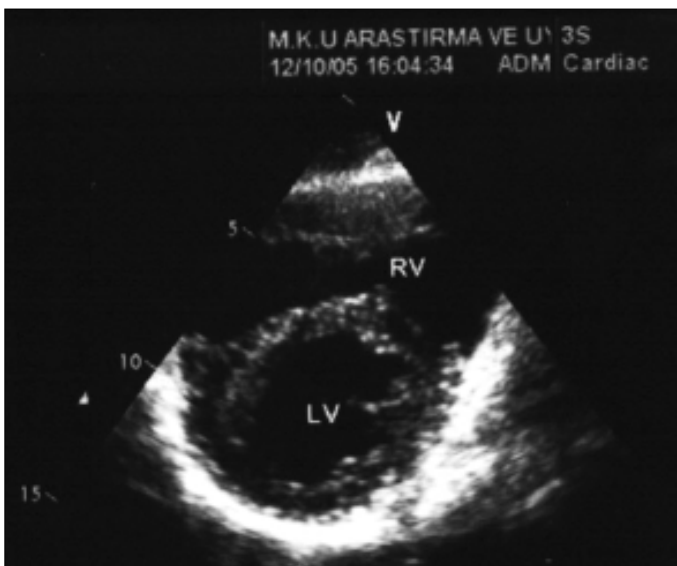


Figure 3. Post-operative interventricular septal thickening (1.0 cm) in parasternal short-axis view

Although right ventricular obstruction in HCM is usually associated with LVOT obstruction, some cases of "isolated" right ventricular obstruction have also been described (5). However, the RV involvement with a significant outflow obstruction is uncommon except relatively mild gradient (5–25 mmHg) in RVOT that may occur in some patients with excessive anterior-septal hypertrophy. Our case had severe RVOT obstruction with excessive anterior-septal hypertrophy (2.8 cm). Disproportional septal ventricular thickening may occur in patients with a variety of congenital heart malformations (3). Hernandez-Reyes et al. (6) reported a case of HCM patient without outflow obstruction with ASH and ostium secundum type septal defect. Recently, Tikanoja et al. (7) have reported muscular VSD in 4 of 8 children with HCM who had mutations in the cardiac β -myosin heavy chain gene. However, we determined perimembranous type of VSD in our case. Asymmetric septal hypertrophy patients with congenital cardiac malformations could have severe RV hypertrophy and RVOT obstruction. The optimal treatment for patients with significant RV disease is unknown. However, medical and surgical therapies have been attempted with variable success.

Echocardiography may have some limitations because of complex three-dimensional structure and anatomic location of right ventricle. Other cardiac imaging methods such as cardiac magnetic resonance imaging and transesophageal echocardiography, may be used.

We suggest that patients with ASH should be carefully evaluated by echocardiography and other methods to detect the RVOT obstruction and the additional congenital cardiac malformations, because it may be more frequent than conventionally deemed.

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