Dilated cardiomyopathy in a patient with quadricuspid aortic valve

Kuadriküspid aort kapağı olan bir hastada dilate kardiyomiyopati gelişimi

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Quadricuspid aortic valve is a rare congenital malformation which used to be discovered incidentally at necropsy, or during aortic valve replacement surgery or aortic angiography until the recent advances in transesophegeal echocardiography. It mostly results in aortic insufficiency, though it may be functionally normal. Congestive heart failure was detected in a 33-year-old male patient with a history of acute rheumatic fever. Transthoracic echocardiography showed dilatation of the left ventricle, thickening and fibrosis of the mitral valve, third degree mitral regurgitation, and a quadricuspid aortic valve. Coronary angiography showed global hypokinesia, aortic and mitral insufficiency. Aortic valve replacement and mitral valve repair were recommended to the patient.

Key words: Aortic valve/abnormalities; aortic valve insufficiency; echocardiography; mitral valve insufficiency; rheumatic fever/complications.

Kuadriküspid aort kapağı oldukça nadir bir doğuştan anomalidir. Önceleri otopsilerde, aort kapak replasmanı sırasında ve aort kökü anjiyografisinde tesadüfen rastlanmakta iken, günümüzde transözofajiyal ekokardiyografideki gelişmeler sayesinde daha erken dönemlerde saptanabilmektedir. Kuadriküspid aort kapağı, fonksiyonel olarak normal olabileceği gibi çoğunlukla aort yetmezliğine neden olmaktadır. Otuz üç yaşında, akut romatizmal ateş öyküsü olan erkek hastada dekompanse sol kalp yetersizliği saptandı. Transtorasik ekokardiyografide sol ventrikül dilatasyonu, mitral kapakta kalınlaşma ve fibrozis, üçüncü derece aort yetersizliği ve kuadriküspid aort kapağı saptandı. Koroner anjiyografide global hipokinezi, aort ve mitral kapak yetersizliği izlendi. Hastaya aort kapak replasmanı ve mitral kapak tamiri önerildi.

Anahtar sözcükler: Aort kapağı/anormallik; aort kapağı yetersizliği; ekokardiyografi; mitral kapağı yetersizliği; romatizmal ateş/komplikasyon.

Quadricuspid aortic valve is a rare congenital malformation, far less common than bicuspid or unicuspid aortic valves.[1] Most of the cases are discovered incidentally at necropsy, during aortic valve replacement, or aortic angiography.[2] Today, these malformations are more likely to be detected before surgery, due to the advances in echocardiography, in particular transesophageal echocardiography.

CASE REPORT

A 33-year-old man who had a history of acute rheumatic fever presented with dyspnea that had developed within the past month. Physical examination revealed increased venous distension, bilateral crepitant rales, S3, systolic and diastolic murmurs at aortic and mesocardiac areas and a systolic murmur at the apex of the heart. There was left bundle branch block in an 12-lead electrocardiogram and chest X-ray showed marked pulmonary congestion. Transthoracic echocardiography revealed left ventricular dilatation, with end-systolic and end-diastolic diameters being 67 mm and 86 mm. Left ventricular systolic function was decreased due to global hypokinesia, ejection fraction was 42% by the modified Simpson's technique. The mitral valve was thick and fibrotic, posterior leaflet motion was limited, and there was diastolic doming of the anterior leaflet. There was third degree mitral regurgitation due to systolic coaptation failure. The aortic valve was quadricuspid, and third degree aortic regurgitation was observed (Fig. 1a, b). The peak systolic gradient was 12 mmHg. He was hospitalized with the diagnosis of decompensated heart failure

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and furosemide infusion was started. After improvement in his clinic situation, coronary angiography was performed which showed global hypokinesia and third degree aortic and mitral insufficiency. No atherosclerotic lesion was observed in the coronary arteries. Cardiac catheterization revealed a pulmonary capillary wedge pressure of 15 mmHg. Aortic valve replacement and mitral valve repair were recommended to the patient.

DISCUSSION

Quadricuspid aortic valve is a rare congenital malformation which is usually detected as an incidental finding at autopsy, during aortic valve replacement or aortic angiography incidentally. Quadricuspid aortic valve is classified into seven types (a to g) according to the size variation of the four cusps.[1,3] Normally, after septation of the embryological arterial trunk, three mesenchymal swellings develop into semilunar leaflets of the aortic and pulmonary trunk in the fourth gestational week.[4,5] In the setting of quadricuspid aortic valve, the formation of the aortic root, sinuses, leaflets, and interleaflet triangles deviate from normal.[6] Unicuspid and bicuspid semilunar valves are more frequently encountered and they are usually incompetent or stenotic.[7] Quadricuspid aortic valves are far less common and they are more frequently seen in the pulmonic valves than the aortic valves.[8] There are very few cases of pentacuspid aortic valves in the literature.[4]

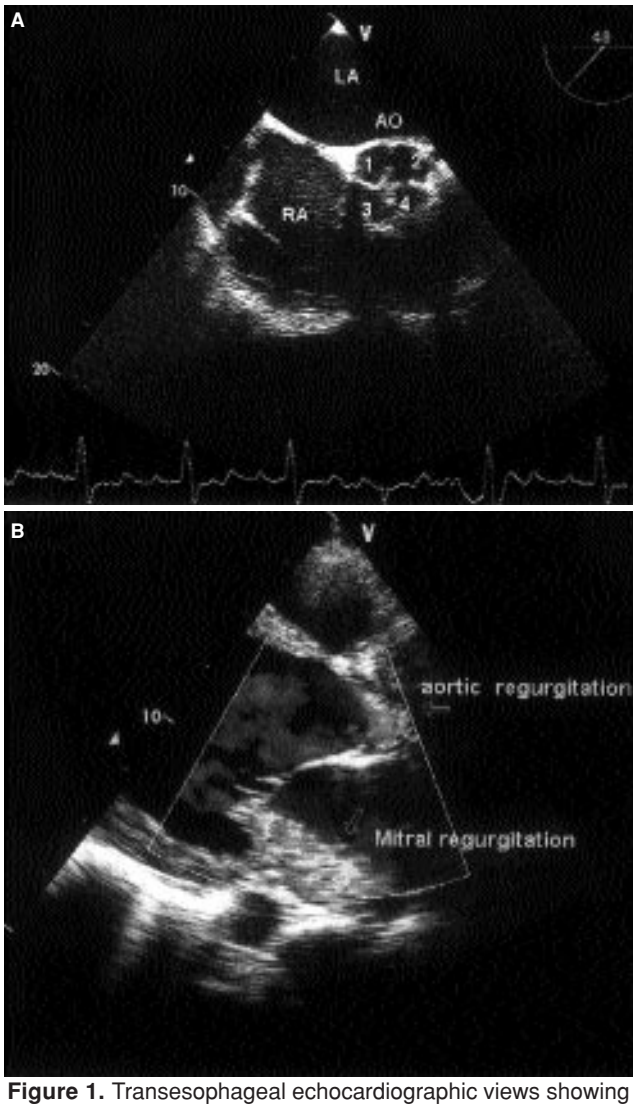

Figure 1. Transesophageal echocardiographic views showing **(A)** the aortic valve with four equal-sized aortic valve cusps, and **(B)** severe mitral and aortic regurgitation.

A series of 60,000 autopsies yielded only two quadricuspid aortic valves (incidence 0.008%).[1] In a retrospective echocardiographic study, eight quadricuspid aortic valves were detected in 60,446 patients (incidence 0.013%).[9] In a literature review by Janssens et al.[2] 70 patients with a quadricuspid aortic valve were assessed. Aortic insufficiency was found in 39 cases (56%), requiring aortic valve replacement in 26 cases; however, valvular stenosis was rare. Eighteen patients were found to be functionally normal. Butany et al.[10] reviewed 247 cases of aortic valves surgically excised over a-year period. Of these, 172 valves (69.6%) were tricuspid, while 74 valves (30.4%) were congenitally abnormal, which included 67 (27.1%) bicuspid valves, six (2.4%) unicommissural valves, and one (0.4%) quadricuspid valve.

There are several reports about other congenital malformations that are associated with a quadricuspid aortic valve. The most frequent abnormalities include coronary osteal or coronary arterial abnormalities.[2,9,11] Also reported are ventricular septal defect, patent ductus arteriosus, subaortic fibromuscular stenosis, mitral valve abnormalities, and obstructive cardiomyopathy.[1,2,12-14]

In our patient, there was severe aortic and mitral insufficiency. The mitral valve was thick and fibrotic, posterior leaflet motion was limited, and there was diastolic doming of the anterior leaflet. Third degree mitral insufficiency was present due to systolic coaptation failure. Considering these, mitral insufficiency was regarded as a sequela of acute rheumatic fever rather than an accompaniment of quadricuspid aortic valve anomaly. In patients with rheumatic carditis, mitral insufficiency is common due to ventricular dilatation or restriction in the motion of the leaflets.[15] During acute rheumatic fever, contractile dysfunction is closely related to the degree of valvu-

lar regurgitation.[16] Chronic pressure and volume overload are related to myocardial damage and decreased contractility.

In our patient, the aortic valve was quadricuspid in nature and all of the four cusps were equal in size. There was severe aortic insufficiency. Severe mitral insufficiency was observed as a sequela of acute rheumatic fever and due to ventricular dilatation caused by severe aortic regurgitation through the quadricuspid aortic valve.

Since medical therapy is very limited for congenital valvular diseases, it is very important to detect these malformations at early ages. Especially in the presence of a congenital valvular malformation, addition of acute rheumatic fever to the clinical situation will worsen the prognosis as seen in this patient. Severe valvular regurgitations resulted in dilated cardiomyopathy. Coronary vessels were normal without any atherosclerotic lesions or congenital abnormalities, excluding ischemic causes of cardiomyopathy.

In conclusion, quadricuspid aortic valve is a very rare malformation which is important to detect due to its severe consequences. In addition, protection of these patients from acute rheumatic fever is another significant issue.

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