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Short Report

Fatih YALÇIN
İrfan SABAH

Marfan Syndrome With Bicuspid Aortic Valve Diagnosed By Transesophageal Echocardiography

Department of Cardiology, EMERGENCY Hospital, Balgat, Ankara-Turkey

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The cadiovascular manifestations of Marfan Syndrome (MS) are usually a result of aortic dissection or rupture of the thoracic aorta or both, or areaused by cardiac failure secondary to progressive aortic regurgitation (1). The diagnostic criteria of MS include ocular manifestations, characteristic skelatal deformities, cardivascular manifestations and family history (2, 3).

In diagnosing MS, even though investigators recently identified a defective FBN 1 gene as cause of this hereditary condition, employing a genetic test a sa diagnostic tool is not partical in the near future (3). This is because of the extreme variability in the FBN 1 gene on chromosome 15. It has been shown (1) that transesophageal echocardiography (TEE) provides rapid diagnostic information in patients with MS with suspected aortic dissection and enhances the assessment of cardiovascular manifestations of this condition. Cystic medial necrosis and bicuspid aortic valves in patients without the overt clinical signs of MS are reported in (4). Using TEE, it has been shown (5) that bicuspid aortic valves are associated with ruptured aortic dissection in the left atrium. Despite many reports in which TEE has been shown to reveal cardiovascular manifestations of MS, the association of MS with bicuspid aortic valves has not been made.

The patient described in this study, a 42 year-old male with a complaint of periodic chest pain, exhibited classical systemic and cardiovascular signs of Marfan syndrome.

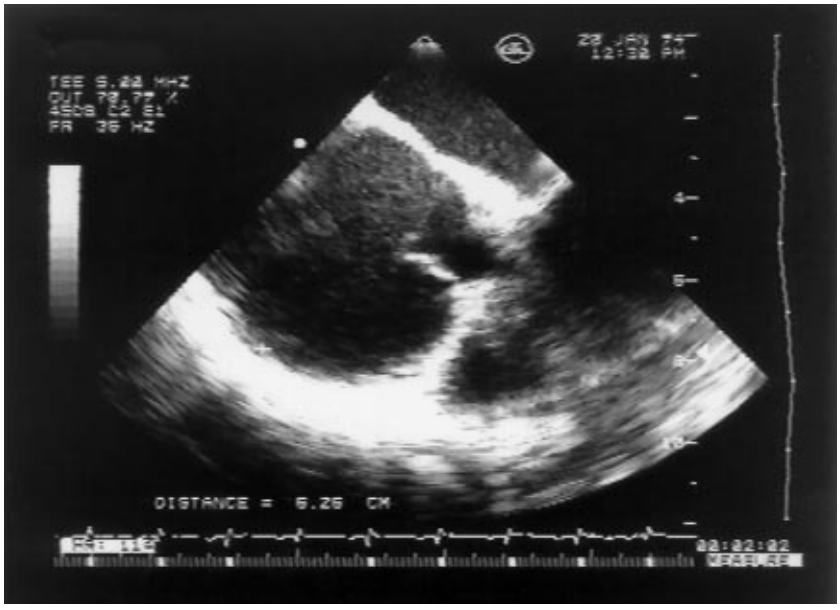

Figure1. Marfan S. with Bicuspid Aortic Valve diagnosed by TEE.

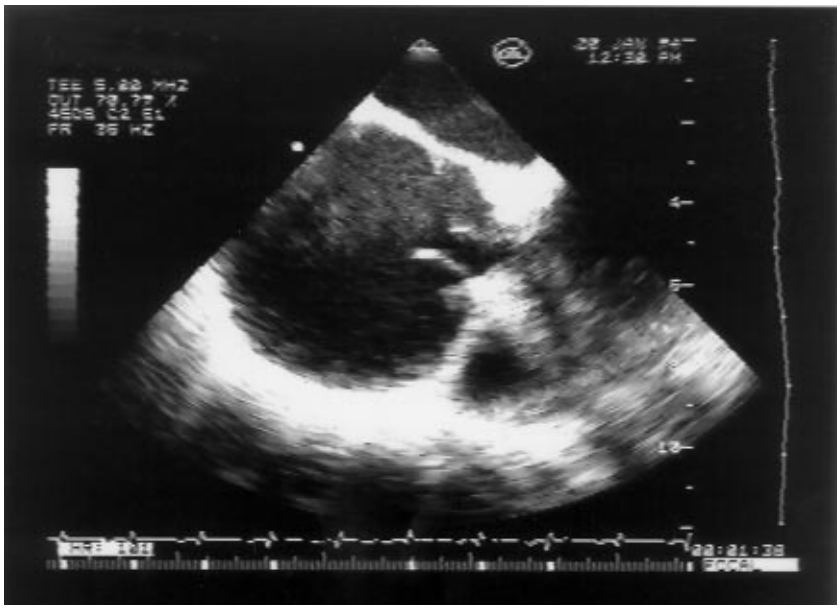

Figure 2. Marfan S. with Bicuspid Aortic Valve diagnosed by TEE.

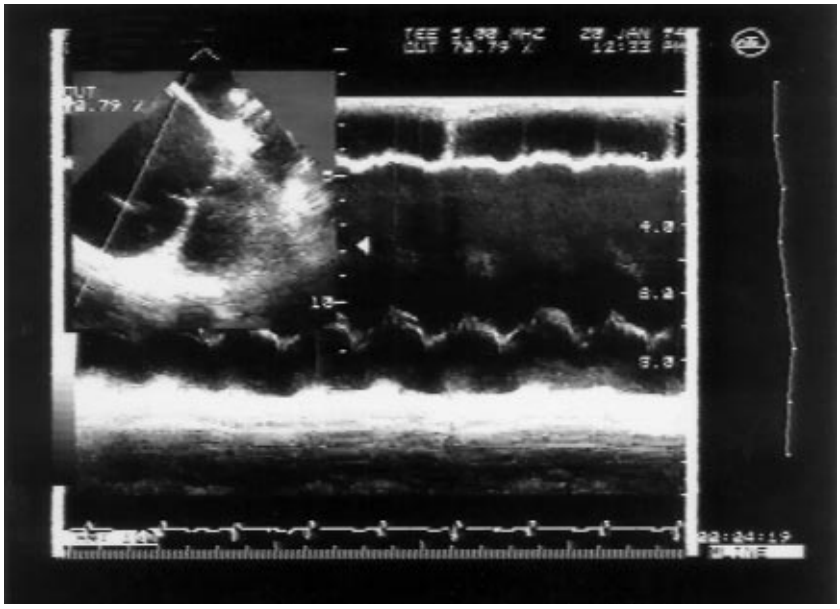

Figure 3. Marfan S. with Bicuspid Aortic Valve diagnosed by TEE.

His chest pain was characterized by a sudden onset and a gradual decrese in severity over several hours and was well localized in the middle of the chest. He described dyspnea and palpitation on exercise. His blood pressure was 110/70 mm Hg, and pulse rate was 86/min He had thorax front wall deformity (pectus excavatum), vertebral colon deformity (thoracic part abnormality) and direct radiography showed an advanced degree of dilatation of the arcus aorta. He reported a family history of early death due to undiagnosed conditions. He exhibited arachnodactyly and reported that his brother also had the same condition. The electrocardiogram did not show any abnormality. However, transthoracic echocardiography revealed aortic dissection with aneurysm and bicuspid aortic valves.

The diagnosis was confirmed by TEE in which aortic root and ascendant aorta dilatation (6.2 cm) with dissection and bicuspid aortic valves were observed (Figure 1). Aortic aneurysm was detected to be associated with bicuspid aortic valve (Figure 2). The evaluation of dissection with m-mode in TEE showed a stagnant appearance in the false lumen (Figure 3). Color flow Doppler mapping displayed a 3˚ aortic regurgitation in the dilated left ventricle that had hypokinetic myocardium.

After determining dissecting aortic aneurysm, the patient was transferred to another hospital for surgery immediately. The patient was operated on soon after diagnosis with graft replacement of the aortic root and normal cardiac function was observed during the postoperative period.

Based on current data on about 450 MS patients at the Cleveland Clinic Foundation, it has been reported recently (3) that, despite new genetic findings, the best way to diagnose MS is to go by the classical clinical manifestations.

In 70-85% of patients with MS, dilatation of the aortic root was observed with combined aortic regurgitation in 20-33% of the cases (6).

The incidence of MS in a previous series of studies (7) noted that aortic dissection varied between 2.6 and 6.3%. The decision made was to operate on the patient because the aneurysm had a dimension of more than 6 cm and was associated with dissection, and the patient was in his forties which is an early mortality period for this group of patients (2). Epperlein (7) has shown that TEE provides important information in the diagnostic approach to patients with aortic dissection and this information cencerns not only the morphology of the aortic dissection but also the underlying pathology of the aorta and aortic valve.

In our case, using TEE enabled the evaluation of the bicuspid aortic valve associated with the aortic aneurysm and dissection and also confirmed the diagnosis provided by transthoracic echocardiography. Furthermore, using TEE facilitated the assessment of the aortic valve, which has been shown (7) to be important in the preoperative planning of the surgical procedure to be employed.

A definitive reason for myocardial hypokinesis could not be investigated during the limited preoperative period and it was thought that frank overload and eccentric left ventricular dilatation due to severe aortic regurgitation might be the basic reasons for decreased cardiac performance.

In conclusion, this article reports a first time demonstration of the diagnosis of Marfan Syndrome associated with bicuspid aortic valves using transesophageal echocardiography. We have shown that a detailed description of the aortic valves can be obtained quickly and reliably with transesophageal echocardiography, which will also provide additional information to assist surgery in terms of evaluation of aortic aneurysm, dissection and aortic regurgitation.

Correspondence author:
Fatih YALÇIN
Tomruk Sok. Gonca Apt. 3/13
A.Ayranci/Ankara

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