

## MARFAN SYNDROME AND QUADRICUSPID AORTIC VALVE

### MARFAN SENDROMU VE KUATRİKÜSPİT AORT

Hüseyin SÜRÜCÜ\*, Sadi ÇİMEN\*\*

#### ABSTRACT

While the major cardiovascular manifestation in Marfan syndrome is a progressive dilatation of the ascending aorta, leading to aortic aneurysm formation and eventually to fatal aortic rupture or dissection, mitral valve prolapse and calcification of the mitral valve annulus, dilatation of the main pulmonary artery may be seen. There was no knowledge about the association of Marfan syndrome and quadricuspid aortic valve. In this case report, we aimed to declare this association between Marfan syndrome and quadricuspid aortic valve.

**Key words:** Marfan syndrome, quadricuspid aortic valve

#### ÖZET

Marfan sendromunun başlıca kardiyovasküler tutulumları arasında progresif çıkan aort dilatasyonu, aorta rüptürü, mitral kapak prolapsuzu ve mitral anuler kalsifikasyon, pulmoner arter dilatasyonu sayılabilir. Ancak Marfan sendromu ile Kuadriküspit aort arasında bir ilişki henüz tanımlanmamıştır. Biz bu olgu sunumumuzda Marfan sendromu ile Kuadriküspit aort arasındaki ilişkiyi vurgulamak istedik.

**Anahtar kelimeler:** Marfan sendromu, kuadriküspit aort.

#### INTRODUCTION

Marfan syndrome (MFS) is a systemic disorder of connective tissue with autosomal dominant inheritance. The diagnosis of MFS is based on the identification of a combination of clinical manifestations in the ocular, musculoskeletal, and cardiovascular organ systems defined in the Ghent Nosology (1). Confirmation of the diagnosis in an individual requires the presence of major clinical manifestations in at least two organ systems associated with involvement of a third organ system. The major cardiovascular manifestation in MFS is a progressive dilatation of the ascending aorta, leading to aortic aneurysm formation and eventually to fatal aortic rupture or dissection. Also, mitral valve prolapse and calcification of the mitral valve annulus, dilatation of the main pulmonary artery may be seen. Recently, De Backer (2) reported that MFS patients present a combination of systolic and diastolic dysfunction that is not related to valvular heart disease. This may be attributed to a primary contractile dysfunction of the myocardium and is likely related to the underlying alterations in the elastic features of the myocardium, resulting from the microfibrillar defect. On the other hand, there was no knowledge about the association of

Marfan syndrome and quadricuspid aortic valve. In this case report, we aimed to declare this association between Marfan syndrome and quadricuspid aortic valve.

A 42-year-old man presented to our hospital with chest pain. Lens ectopia and ocular myopia was present. Marfanoid habitus (arachnodactyly, scoliosis and joint hypermobility) was also prominent. In his cardiac examination findings, there was no abnormality (there was no systolic and/or diastolic murmur). His ECG was consistent with incomplete right bundle branch block (Figure 1). There was no abnormality in his laboratory findings. His treadmill exercise test way found negative. Echocardiography was performed. The mitral valve leaflets had elongated and redundant appearance. The sinuses of Valsalva were measured as dilated (aortic root: 43 mm), but we found a striking finding at parasternal short axis examination. His aortic valve was quadricuspid (Figure 2). Mitral or aortic regurgitation was not found in trans-thoracic echocardiography (TTE). Because quadricuspid aortic valve was seen very clearly and proximal aorta was not aneurysmatic in TTE, trans-esophageal echocardiography or another cardiac imaging (MRI or CT) was not required.

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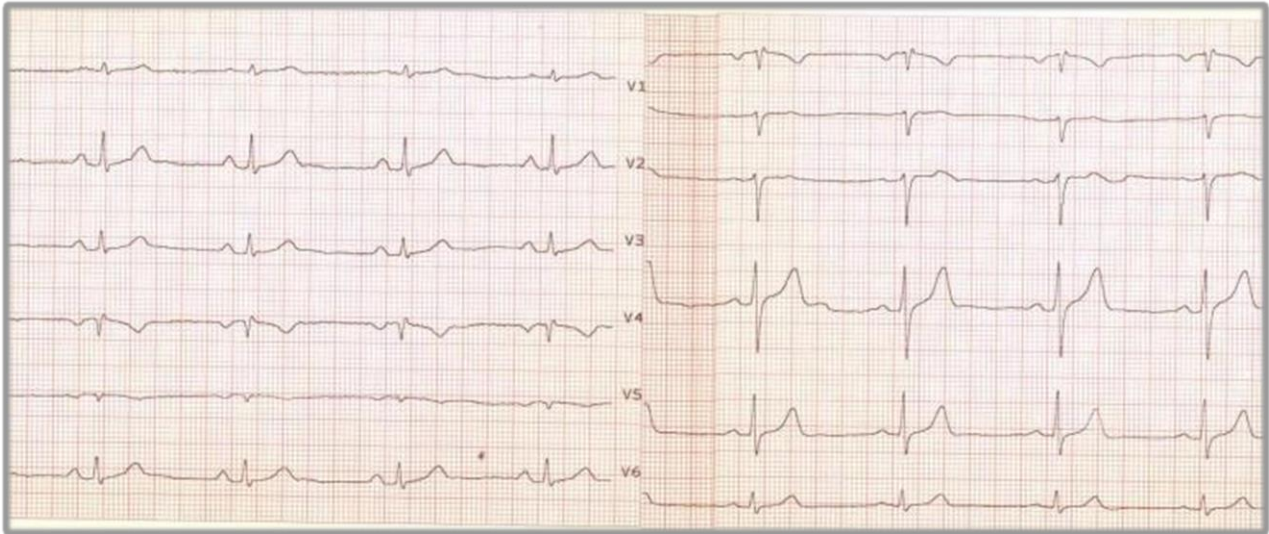
Private Avcılar Anadolu Hospital, \* Department of Cardiology, \*\* Department of Internal Medicine, İstanbul Turkey.

(İletişim kurulacak yazar: : huseyinsurucu@hotmail.com)

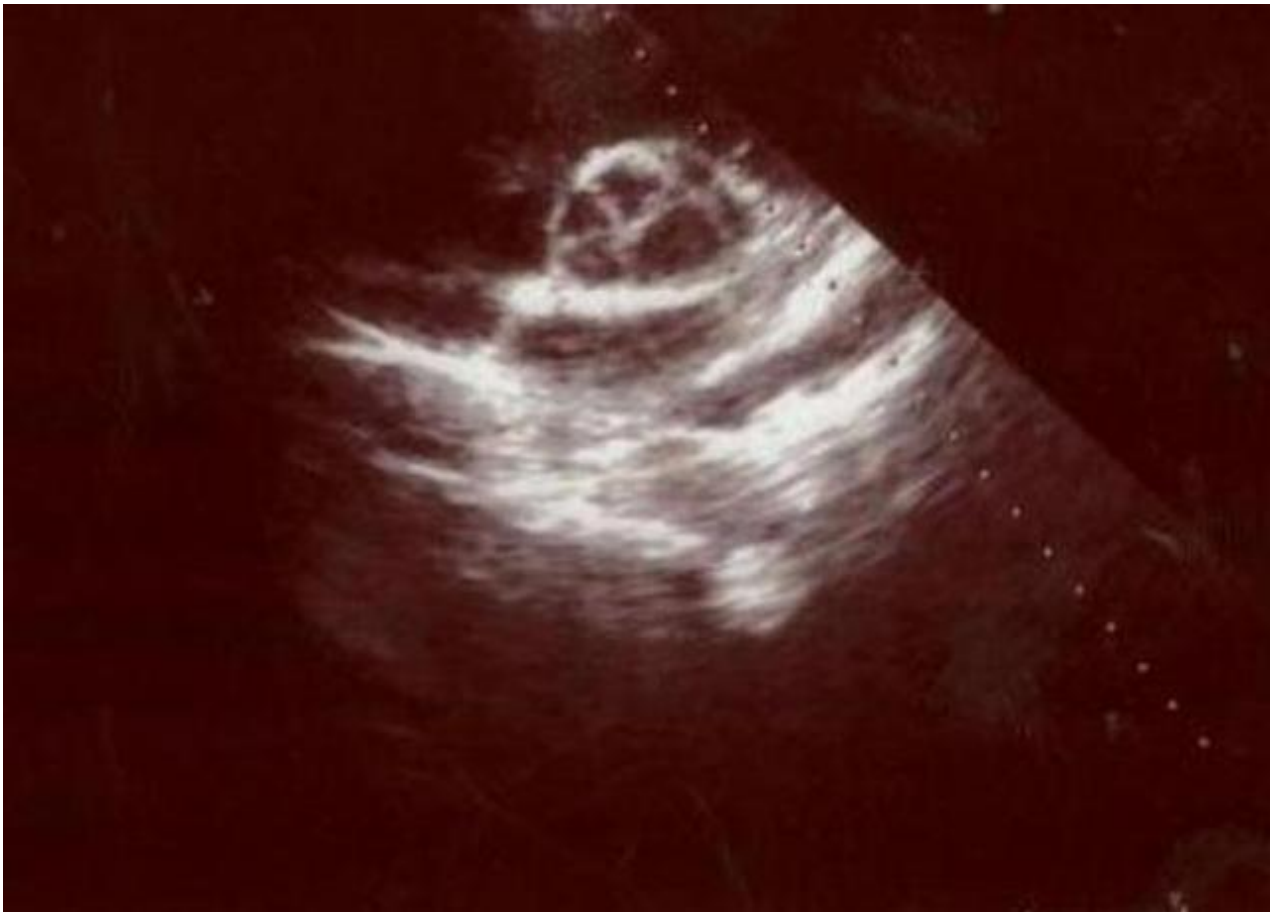
## ***Marfan syndrome and quadricuspid aortic valve***

When the literature was reviewed, there was no association between Marfan syndrome and quadricuspid

aortic valve. We think that our case may be the first reported quadricuspid aortic valve in Marfan syndrome.



**Figure 1: Patient's electrocardiography**



**Figure 2: Patient's quadricuspid aortic valve.**

### **REFERENCES**

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