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Isolated infrarenal abdominal aorta aneurysm in a 42-year-old patient with Marfan's syndrome: Case report

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Abstract

Marfan's syndrome is an autosomal dominant disorder of connective tissue characterized by a large number of possible mutations and by heterogeneity of clinical presentation primarily in skeletal, ocular and cardiovascular organ systems. Cardiovascular complications of the disease are responsible for high mortality. The case of a 42-year-old patient with a progressive advanced abdominal aorta dilatation visualized on computed tomography images is presented. Pathogenesis, diagnosis and management of patients with Marfan's syndrome are also discussed.

Keywords

Marfan's syndrome, abdominal aortic aneurysm, angio-CT, surgery

Introduction

Marfan's syndrome is an autosomal dominant disorder of connective tissue caused by mutations in the *FBN1* gene on chromosome 15q21, which are responsible for creation of a wrong fibrillin-1.[1] The disease is characterized by a large number of possible mutations and by heterogeneity of clinical presentation primarily in skeletal, ocular and cardiovascular organ systems. Cardiovascular complications are mainly represented by mitral valve prolapse and regurgitation (60%–80%), but aortic aneurysm, especially aortic root (66.7% of cardiovascular complications), is the most common cause of morbidity and mortality. Aneurysms of the abdominal aorta (AAA) remain a rare complication; so we consider it appropriate to report a new observation studied recently.

Case report

A 42-year-old patient was admitted to our department with abdominal mass beating. The patient had been diagnosed with Marfan's syndrome on the basis of a typical clinical picture: patient slender, flat feet (Figure 1) and positive signs of laxity including Steinberg sign and wrist sign (Figure 2). Abdominal examination found a per umbilical mass, painless, swinging, expansive character and a positive sign Debakey. Duplex ultrasonography and computed tomography (CT) scan (angio-CT scan) demonstrated an aneurysm of infrarenal aorta with transversal diameters of 60.1 mm × 64.9 mm (Figures 3 and 4). No complications of the existing aneurysm were observed. We realized an echocardiography and a fundus which were normal. The aneurysm was surgically treated under general anaesthesia. It was then dissected and exposed by the transperitoneal approach. We realized an aneurysmorhaphy and an aorto-biiliaque bypass using a prosthesis. No per-operative complications were encountered. At 1-year follow-up, the patient was asymptomatic without any complications.

Discussion

The diagnosis of Marfan's syndrome is primarily based on clinical manifestations, which have an extraordinary degree of variation. Chromosomal analyses of gene defects on chromosome 15 and histological investigations of cystic media necrosis (Erdheim Gsell) continue to be less reliable.[2,3] Considering the life expectancy of these patients, an early diagnosis is extremely important to prevent typical complications. Cardiovascular complications, which in 30%–60% of the cases lead to a reduction in life expectancy, are of special relevance.

The most common vascular manifestations of the disease include mitral valve prolapse and regurgitation. Aortic dilatation, however, is the most common cause of morbidity and

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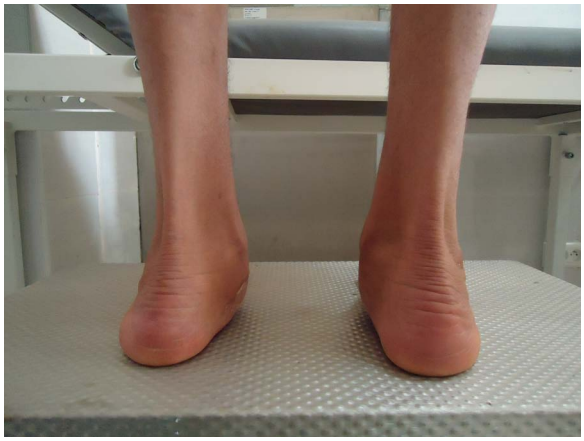

Figure 1. Flat feet.

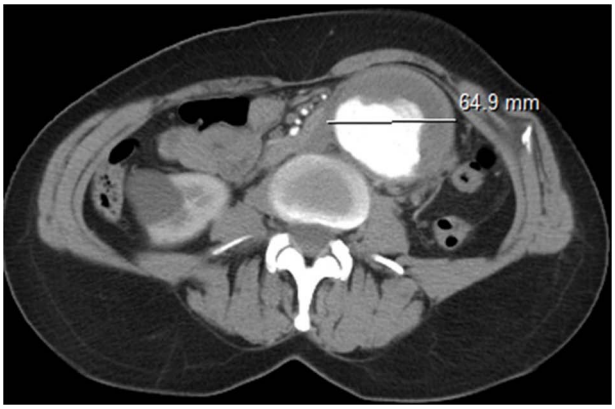

Figure 3. Axial angio-CT scan showing the aneurysm.
CT: computed tomography.

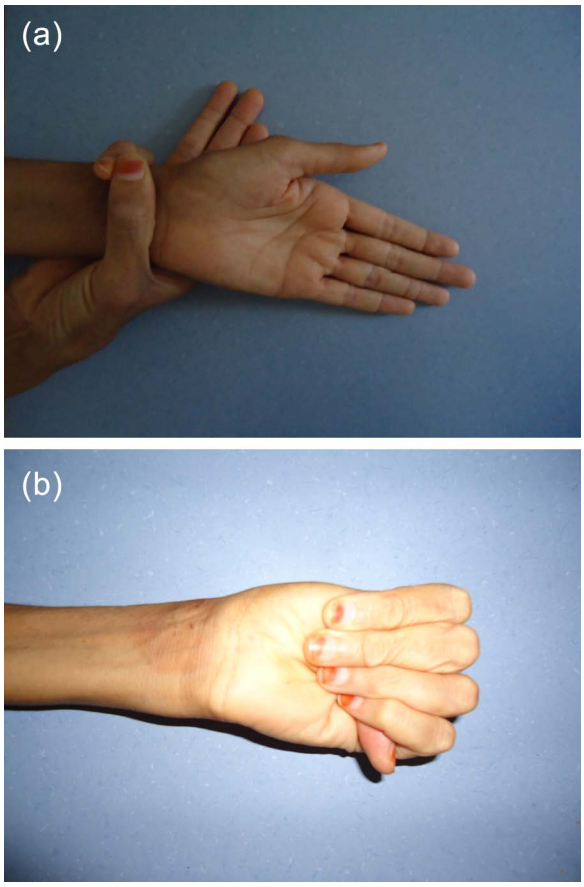

Figure 2. (a) Wrist sign and (b) Steinberg sign.

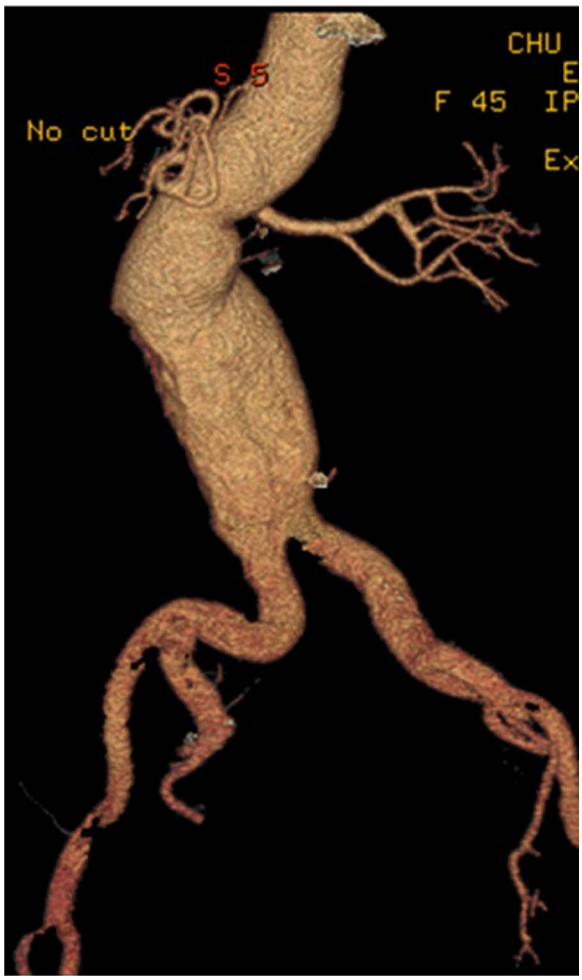

Figure 4. Reconstructed angio-CT scan showing the aneurysm.
CT: computed tomography.

mortality. Aortic dilatation usually occurs primarily in the sinuses of Valsalva and the aortic root.[3] Then the ascending aorta becomes involved, followed by the arch and descending thoracic aorta, all of which might occur in patients at an early age.[4] In presented case, the patient did not have a thoracic aorta aneurysm, but developed earlier a huge AAA.

AAA occurred in relatively young patients with Marfan's syndrome. A small aneurysm had a great percentage for rupture or dissection due to the fragility of the aortic wall, especially in the tunica media.[5]

Imaging methods, such as CT or magnetic resonance imaging (MRI), are therefore the methods of choice for

assessing the risk and monitoring patients with Marfan's syndrome. The AAAs seen in Marfan's syndrome also rarely show intimal calcification or atherosclerotic thrombosis, and they appear as a cystic or fusiform dilatation of the aorta.[6]

The treatment of choice is an on-lay prosthetic graft. The aneurismal sac is usually closed over the graft in order to prevent the formation of aortoduodenal fistula and to limit the infection. The main stay of treatment in symptomatic patients is excision and repair with tube graft prosthesis. In asymptomatic aneurysm with a diameter >5 cm, repair should be proposed because of the high risk of rupture and high rate of mortality.[7] The place of non-surgical treatment is restricted to the use of beta blockers to slow down the rate of aortic dilatation.[8]

Despite the recent and vast expansion of endovascular aneurismal repair for atherosclerotic AAA, there is no established consensus on endovascular repair for Marfan-related AAA.[5]

Declaration of conflicting interests

The authors declare that there is no conflict of interest.

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