

Huge isolate abdominal aorta aneurysm in a 24-year-old patient with Marfan syndrome in CT imaging in view of recent literature

Ewa Kurys-Denis, Witold Krupski

II Department of Radiology, Medical University, Lublin, Poland

Abstract: The Marfan 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 young patient with a progressive advanced abdominal aorta dilatation visualized on CT images is presented. Pathogenesis, diagnosis and management of patients with Marfan syndrome are also discussed.

Key words: Marfan syndrome, abdominal aneurysm, CT

INTRODUCTION

The Marfan syndrome is an autosomal dominant disorder of connective tissue caused by mutations in the *FBNI* gene on chromosome 15q21 which are responsible for creation of a wrong fibrillin-1, the major constituent of extracellular microfibrils. 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 of the disease are responsible mainly for its adverse effect on mortality. Prognosis is mostly determined by progressive dilatation of the aorta, potentially leading to aortic dissection and death at a young age. Medical management involving beta-adrenergic blocking agents is recommended from a very young age, until severe dilatation requiring surgery is present. The degree of aortic dilatation has been correlated with the risk of aortic rupture. Therefore, early diagnosis and close follow-up of patients with Marfan syndrome is important to detect progression of aortic dilatation. Computed tomography (CT) with its multiple planar reconstructions (MPR) and volume rendering technique (VRT) images is a very good tool enabling precise calculation of aortic dimensions and detecting possible aortic complications.

CASE REPORT

A 24-year-old patient was admitted to hospital with sudden abdominal pain. A suggestion of possible aneurismal complications was made. He was therefore referred to our department for vascular CT examination and an exact assessment of the patient's aorta status.

The patient had been earlier diagnosed with Marfan syndrome on the basis of a typical clinical picture. CT examination of the thoracic, abdominal aorta and iliac arteries was performed in vascular protocol with a Siemens Somatom

Emotion CT scanner, in axial 3 mm slices / pitch 1.5. The scanning was performed before administering the contrast agent, after which enhanced examination was performed using an automatic syringe; 120 ml of contrast agent was injected in 2 phases: in the first phase, which lasted 8 seconds, 4 ml per sec, and the second phase – 2.5 ml per sec. The scanning was automatically started when peak enhancement inside the lumen of the examined aorta was reached. The examination showed a huge abdominal aorta aneurysm, measuring up to 10 cm in diameter, with the presence of a circumferential large thrombus. (Figure 1, 2). The aneurysm involved the entire aorta in its abdominal part, starting just beneath the diaphragm. There was no aorta dilatation in its thoracic part (Figure 3). The abdominal aneurysm extended onto both common iliac arteries and strongly compressed the neighbouring abdominal structures, especially the spleen and renal vessels (Figure 4). For better visualization, all arteries were assessed in the maximum intensity projection (MIP) and on 3D images, created using the Volume Rendering Technique (VRT) (Figure 5, 6). No complications of the existing aneurysm were observed.

DISCUSSION

The Marfan syndrome is an under-recognized inheritable connective tissue disorder, affecting mainly the cardiovascular system, eyes and skeleton. Additionally, the skin, integument, lung, muscle, adipose tissue and dura can also be affected. The incidence of the disease is estimated at 2-3 per 10,000 individuals [1]. It is caused by a mutation in the gene *FBNI* which encodes the fibrillin, and more than 500 types of mutation have already been identified. All are unique to the affected person or family [2]. In spite of this, the diagnosis of the Marfan syndrome is still primarily clinical. The gold standard for the diagnosis of Marfan syndrome are the Ghent criteria, established in 1996 by a team of prominent geneticists [3]. Major criteria in at least 2 organ systems and one minor criterion are required for a diagnosis. One of the major criteria of the Marfan syndrome is dural ectasia, in which CT plays an important role [4]. Our case has a positive family history fulfilling the Ghent criteria.

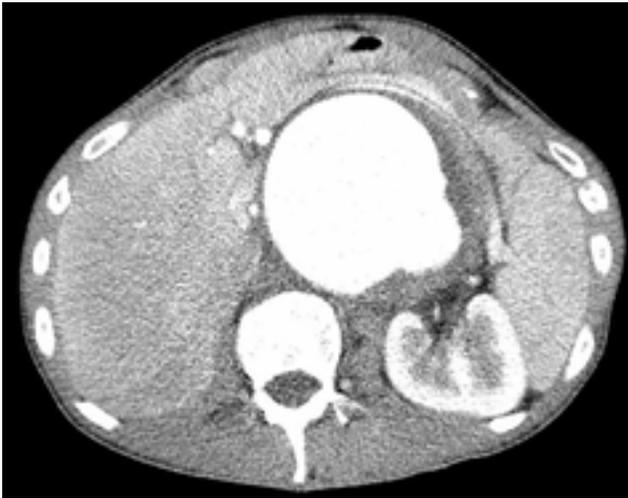


Figure 1 Huge abdominal aorta aneurysm of 10 cm diameter, with a circumferential large thrombus, starting just underneath the diaphragm.



Figure 4 Aorta aneurysm at the level of renal arteries. Note the big effect of compression of renal and spleen vessels.

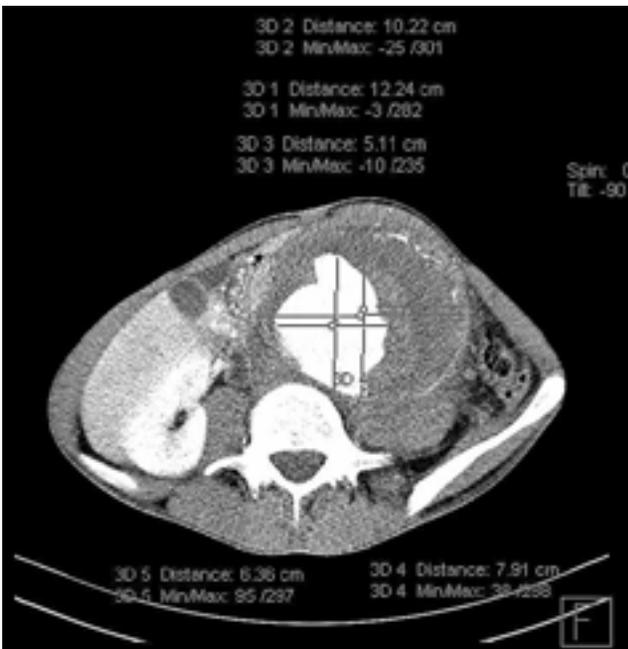


Figure 2 The abdominal aneurysm at the level of aorta bifurcation, extending to both common iliac arteries.

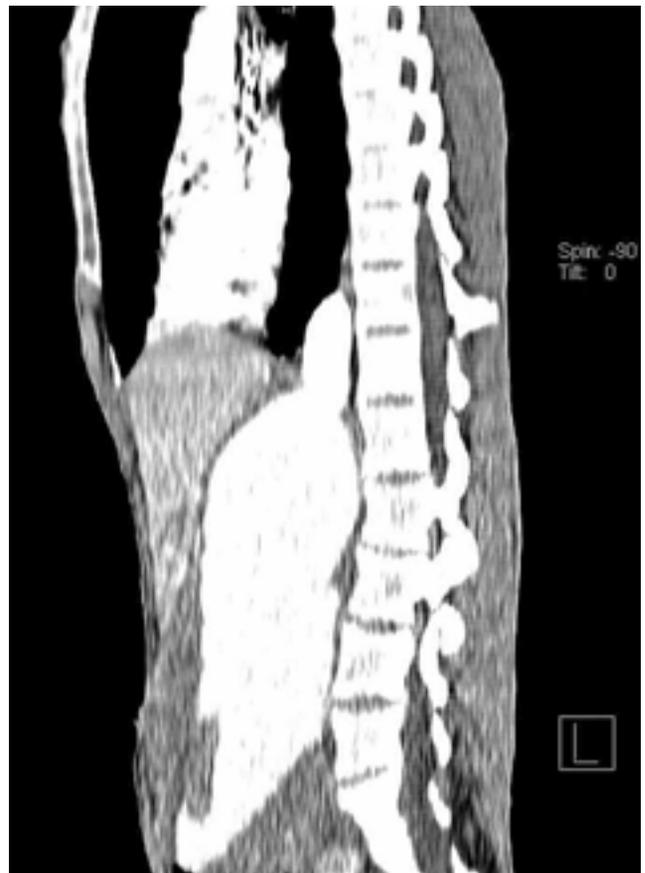


Figure 5 The abdominal aneurysm assessed in maximum intensity projection (MIP).

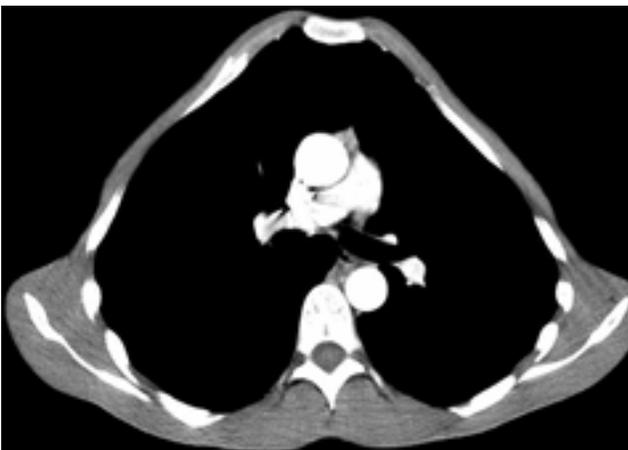


Figure 3 Axial image showing no dilatation of the thoracic aorta.

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 mortality. Aortic dilatation usually occurs primarily in the sinuses of Valsalva and the aortic root [5]. 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 [9]. Involvement of the abdominal aorta is



Figure 6 The abdominal and iliac aneurysm visualized on 3D images, created using Volume Rendering Technique (VRT).

a relatively uncommon early manifestation. In presented case, the patient did not have a thoracic aorta aneurysm, but developed earlier a huge abdominal aorta aneurysm. In patients with aortic aneurysms not associated with the Marfan syndrome the degree of aortic dilatation has been well correlated with aortic rupture [6]. However, in patients displaying the symptoms of the Marfan syndrome, the risk of aortic rupture or dissection has been proved to depend not solely on the degree of dilatation, but also to be related to the length of the affected segment in such a way that aortic dilatation limited to the sinuses of Valsalva carries a less malignant prognosis than aortic dilatation extending to the aortic arch. The risk of aortic rupture in patients with the Marfan syndrome has also been strongly associated with a family history of severe cardiovascular disease [7]. Recent studies by Nollen et al indicate that both aortic diameter and aortic distensibility are independent predictors of progressive aorta dilatation, the diameter being a better predictor of the abdominal aorta dilatation, and the distensibility being the better predictor of the thoracic aorta dilatation [10]. Imaging methods, such as CT or MRI, are therefore the methods of choice for assessing the risk and monitoring patients with the Marfan syndrome.

Recently, increased transforming growth factor beta (TGF) signaling has been proved to take part in the molecular pathogenesis of fibrillin-1-deficient mice, and indirectly cause aneurysm formation [1]. This revolutionary finding enables a future attractive target to counteract aneurysm progression in Marfan syndrome using pharmacological means of therapy.

At present, management of cardiovascular disease in patients with the Marfan syndrome consists primarily of therapy with beta-adrenergic receptor antagonists, regular follow-up to quantify progression of aortic dilatation and to exclude any aortic aneurysm complications. CT plays an important role in such cases. Identification of patients with high risk of dissection or aortic rupture is crucial for planning the appropriate intervention [8, 12]. Nowadays, aortic valve-sparing technique is more often used in aorta root dilatation or aortic dissection type A in patients with Marfan syndrome. Recent studies assessing the results of this operation show an excellent early outcome, favorable long-term results, and acceptable durability of the re-implanted valve [11].

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