Case Report

An Unusual Case of Large Ascending Aortic Aneurysm with Severe Aortic Insufficiency

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Abstract

The etiology of ascending aortic aneurysms includes genetic, degenerative, inflammatory and infectious causes. We present a case of a 49 year-old male patient without any known cardiovascular risk factors, who presented mainly with fatigue of increasing severity. The physical examination revealed a grade IV decrescendo, blowing aortic diastolic murmur, as well as an Austin Flint murmur and water hammer peripheral pulses. Imaging methods (echocardiography and computed tomography angiography) showed a large (8.6 cm) ascending aortic aneurysm with corresponding severe aortic insufficiency. In this context, he underwent surgical implantation of an aortic tube graft and mechanical prosthesis in aortic position, with good final result. Surprisingly, the histopathological diagnosis of the surgical specimen was compatible with atherosclerosis.

ABBREVIATIONS

BNP: Blood Natriuretic Peptide; CTA: Computed Tomography Angiography; MRA: Magnetic Resonance Angiography; TEE: Transesophageal Echocardiography; TTE: Transthoracic Echocardiography

INTRODUCTION

Thoracic aortic aneurysms have an incidence of 10/100 000 person-years and approximately 60% affect the ascending aorta [1]. The etiology of ascending aortic aneurysms includes genetic (such as Marfan, Loeys-Dietz or Ehlers-Danlos syndrome; bicuspid aortic valve), degenerative (atherosclerotic), inflammatory (for instance, giant cell arteritis) and infectious causes (Treponema pallidum, Staphylococcus aureus, Salmonella spp., among others) [1]. Nevertheless, atherosclerosis is the main cause of descending but not ascending aortic aneurysms [1]. The clinical presentation is variable, with most of the patients remaining asymptomatic for a long period of time. The symptoms are usually related to aortic insufficiency, heart failure, local compression of adjacent structures (cough, dyspnea, odynophagia or dysphagia, hoarseness due to left laryngeal nerve palsy) or even systemic embolization [1,2]. However, the first manifestation may correspond to the most feared complications: dissection (acute deep, aching or throbbing chest pain that can spread to the back, buttocks, groin or legs) and rupture (that can lead to sudden death) [1,2]. According to the Global Burden Disease 2010 project, the overall global death rate from aortic aneurysms and aortic dissection increased from 2.49 per 100 000 to 2.78 per 100 000 inhabitants between 1990 and 2010, with higher rates for men [2]. The main predictors of rupture are aneurysm size >6.5 cm and its expansion rate >5.5 mm per year, but and history of bicuspid aortic valve or Marfan syndrome are also determining factors of expansion rate [3,4]. Baseline laboratory assessment includes cardiovascular risk factors and albeit laboratory testing plays a minor role in the diagnosis of acute aortic diseases, it is useful for differential diagnoses [2].

The aorta is a complex geometric structure and, consequently, there are some limitations inherent to all imaging techniques (TTE, TEE, CTA or MRA). No imaging modality has perfect resolution and the precise depiction of the aortic walls depends on whether appropriate electrocardiogram (ECG) gating is employed [2,5].

In this regard, it is recommended that maximum aneurysm diameter be measured perpendicular to the centerline of the vessel with three-dimensional (3D) reconstructed CTA scan images whenever possible [2]. In fact, CT plays a central role in the diagnosis, risk stratification and management of aortic aneurysms [2]. Regarding treatment, aneurysms are repaired when the risk of rupture or dissection exceeds the risk of repair. The size thresholds for repair are determined by the
underlying etiology of the aneurysm (usually >55 mm), with lower thresholds for those with genetic aortopathies (45 mm for Marfan’s syndrome and 50 mm for bicuspid aortic valve) [1,5-7].

CASE PRESENTATION

We present the case of a 49 year-old male patient who lived in Cape Verde, Africa and worked in the construction field. This patient had no relevant past medical story and no identifiable cardiovascular risk factors (arterial hypertension, smoking, hyperlipidemia, diabetes, family history of early cardiovascular disease) what so ever. Nevertheless, he started complaining mainly of progressively worsening fatigue and sporadically some chest discomfort (never described as pain), in the past few months. The patient was initially observed in his homeland and the physical examination revealed a grade IV decrescendo, blowing aortic diastolic murmur, as well as an Austin Flint murmur and water hammer peripheral pulses, suggestive of aortic insufficiency. After a brief workup, the patient was evacuated to Portugal, in order to investigate more thoroughly his condition and decide the best treatment. Regarding laboratory tests, the main results are depicted in Table, with only a slight elevation of BNP levels as a remarkable abnormality. The first imaging exam performed was a chest X-ray that showed both enlarged cardiac silhouette and aortic notch (Figure 1).

The TTE, using both multi planar and 3D acquisition (Figure 2a), showed a severe aortic insufficiency (tricuspid valve; vena contracta = 8.3 mm, pressure half-time = 234 ms (Figure 2b), effective regurgitant orifice area = 0.9 cm² and regurgitant volume = 164 mL (Figure 2c)), secondary to a large ascending aortic aneurysm (Figure 2d), with the following 2D measures: aortic ring = 4.1 cm, Valsalva sinuses = 5.18 cm, sino-tubular junction = 7.1 cm and ascending aorta = 8.7 cm. The left ventricle was enlarged, with ejection fraction (calculated by Simpson’s method and using 3D software) of 41% and global longitudinal strain of -14.4% (Figure 3). Subsequently, the patient had a CTA (with 3D reconstruction) that confirmed the large ascending aortic aneurysm with 8.6 cm (Figures 4a, 4b and 4c). Additionally, it is important to underline that the calcium score was zero, there were no identifiable coronary lesions and there were no other aneurysms identified.

In this context, the patient underwent surgical resection of the aneurysm (Figure 5a), with implantation of an aortic tube graft Jotec® # 30 (Figure 5b) and mechanical prosthesis in aortic position St. Jude® # 25 (Figure 5c), with good final result. Finally,
DISCUSSION

Ascending aortic aneurysms may present with symptoms related to aortic insufficiency, heart failure, local compression of adjacent structures or even systemic embolization [1-2]. However, considering that most aneurysms remain silent for a long period of time, harboring as they grow an increased risk of rupture and sudden cardiac death, it is extremely important their timely diagnosis (with an imaging exam, preferably CTA with 3D reconstruction) and adequate treatment (surgery when indicated) [2]. The size thresholds for surgical repair are determined by the underlying etiology of the aneurysm, with lower thresholds for those with genetic aortopathies [5-7]. In younger people, the most common causes of ascending aortic aneurysms are genetic (especially Marfan syndrome), but with time atherosclerosis becomes more prevalent [6]. On the contrary, atherosclerosis is the main cause of descending aortic aneurysms [1]. This case illustrates a large ascending aortic aneurysm with secondary severe aortic insufficiency and subsequent dilation of the left ventricle with discrete systolic dysfunction, extensively studied both by TTE and CTA. The surgical indication

the histopathological diagnosis of the surgical specimen was compatible with atherosclerosis (disruption of elastic fibers and smooth muscle cells that were partially replaced by extracellular matrix and lipids).
was formerly the main cause of ascending aortic aneurysms, but nowadays, in developed countries, it is much less common following the introduction of effective antibiotics [8]. On the opposite, in developing countries such as Cape Verde, infectious causes remain an important issue. This etiology was, indeed, excluded both by laboratory testing and histopathological exam. Nevertheless, the histopathological exam also excluded connective tissue disorders (CTD), like Marfan syndrome, that typically include cystic medial degeneration (characterized by fragmentation of elastic fibers, smooth muscle cell loss, increase in collagen deposition and replacement with interstitial cyst of mucoid appearing basophilic-staining ground substance) [1]. Considering the histological report and the absence of phenol typical manifestations characteristic of Marfan syndrome and other CTD, we decided not to perform any genetic testing. Despite the absence of known cardiovascular risk factors and of macroscopic plaques according to CTA, the final diagnosis of atherosclerotic origin has been made based on the histopathological exam. On the one hand, the pathogenesis of atherosclerosis multifactorial, involving genetic predisposition and risk factors (such as dyslipidemia, smoking, diabetes, systemic hypertension) that accelerate this process [1]. Nonetheless, there may be other potential risk factors contributing to the development of atherosclerosis, as recently suggested, like trimethylamine (present in many dietary compounds: meat, milk and other foods of animal origin) [9]. On the other hand, histologic examination shows considerable distinction between occlusive atherosclerotic disease - such as coronary artery disease (CAD) – and aneurysmal disease [1], which means that the absence of CAD does not exclude the possibility of an aneurysm of atherosclerotic cause.

In conclusion, this case illustrates a large ascending aortic aneurysm with severe aortic insufficiency that turned out to be of atherosclerotic origin, against all odds, probably due to a genetically determined predisposition and/or an unrecognized vascular risk factor. To the best of our knowledge, this is the first case report published with the finding of such a large ascending aortic aneurysm due to atherosclerosis in a patient without any known cardiovascular risk factors.

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REFERENCES