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Annulo-Ectatic Aortic Disease of Marfanoid Origin: A Case Report with Literature Review.

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A B S T R A C T

Under the generic term annuloaortic ectasia hide multiple pathologies with close phenotypic expression involving dilatation of the aortic root and the aortic annulus. In most cases, the etiology is not found and remains unclear. Hereditary diseases of connective tissue such as Marfan syndrome, inflammatory and rheumatic aortitis or syphilis mark the rest of etiologies. The development of these aneurysms is associated with high mortality, following the physic laws of and evolutes towards dilatation is common. Echocardiography and spiral thoracic computed tomography (CT) are key examinations to assess the diameter of the ascending aorta and its progression over time. Medical treatment with β-blockers and follow-up with echocardiography once or twice a year are recommended for asymptomatic patients. If the size of the aneurysm is the main element to determinate the timing of surgery, other elements, such as underlying etiology, aortic insuffi ciency, growth rate of the aneurysm or nature of the bicuspid aortic valve should be assessed carefully.

Keywords: Aortic aneurysm - Aortic surgery - Aortic root

CASE REPORT

This is a 37-year-old man with no cardiovascular risk factors or significant medical history, who has been experiencing recurrent episodes of chest pain for the past two months.

Clinical Examination:

The patient was found to be eupneic at rest. His blood pressure was 140/60 mm Hg, and his peripheral pulses were full and bounding. Cardiac auscultation revealed a diastolic murmur of aortic insufficiency graded 5/6 at the aortic area.

Electrocardiogram:

The electrocardiogram (ECG) showed left ventricular hypertrophy.

Chest X-ray:

The chest radiograph revealed mediastinal widening with a cardiothoracic ratio of 0.4.

Transthoracic Echocardiogram: (figure1)

The transthoracic echocardiography revealed a dilated left ventricle with slightly hypertrophied walls, hyperkinetic movement, and systolic dysfunction with an ejection fraction estimated at 44%. There was severe aortic insufficiency with aneurysmal dilation of the aortic root and obliteration of the sinotubular junction (Left ventricular end-diastolic diameter: 31mm, Sinus of Valsalva: 73mm, Ascending Aorta: 75mm) and severe aortic regurgitation (Effective regurgitant orifice: 40mm², Regurgitant volume: 50ml, Vena contracta: 10mm, Diastolic flow velocity: 0.4m/s).

Aortic CT Angiography: (figure2)

The aortic CT scan revealed a predominantly aneurysmal dilation in segment I of the ascending aorta, without any signs of complications.

However, an etiological workup was initiated, with a biological assessment showing no abnormalities. The cerebral and abdominal CT angiography, as well as the Doppler ultrasound of the lower limbs, did not reveal any other aneurysmal locations.

Surgical intervention was indicated due to the severity of the aneurysm and the aortic regurgitation, in order to prevent the risk of rupture or dissection. The surgical procedure involved the complete replacement of the ascending aorta while preserving the native aortic valve, using the Tirone David technique under extracorporeal circulation, with uncomplicated postoperative recovery.

DISCUSSION

Aortic aneurysm ranks as the 13th leading cause of mortality in developed countries [1,2]. The incidence of thoracic aortic aneurysms is approximately 4.5 cases per 100,000 individuals [1,3].

Supravalvular aneurysms are less common compared to those of the aortic root and predominantly affect men. The average age for these aneurysms ranges from 59 to 69 years. Conversely, aortic root aneurysms occur in younger individuals, typically between the ages of 30 and 50. Aortic aneurysms are degenerative abnormalities characterized by three main processes: proteolysis, leading to the degradation of the extracellular matrix, the progressive loss of smooth muscle cells in the media, and their replacement by areas of mucoid degeneration, rich in vacuoles and sulfated polysaccharides.

Aortic root aneurysms, often classified under annulo-ectasic aortic diseases, can be either idiopathic or associated with connective tissue diseases such as Marfan syndrome, Ehlers-Danlos syndrome, or bicuspid aortic valve anomalies [1,3].

Approximately 20% of patients with Marfan syndrome require surgery for an aortic root aneurysm. A significant association has also been observed between bicuspid aortic valve disease and aortic dissection, with a tenfold increased risk compared to the general population. Most ascending aortic aneurysms remain asymptomatic for long periods. The diagnosis is often made incidentally during chest X-rays or other imaging modalities, such as echocardiography, performed for concomitant cardiac pathology or as part of a routine screening in individuals with a family history of diseases like Marfan syndrome.

The onset of acute symptoms, particularly a tearing pain, is highly suggestive of imminent rupture or dissection and is observed in approximately 75% of patients. Chronic pain may result from compression of the sternum and is the first symptom in 25 to 75% of patients.

Other symptoms may arise from the pressure exerted by the aorta on adjacent structures, including cough and hemoptysis, dysphagia, dysphonia, superior vena cava syndrome, and more rarely, fistulas with the right atrium or superior vena cava [1].

Clinical examination provides little specific information for diagnosing an aortic aneurysm. An aortic root aneurysm is frequently associated with aortic insufficiency, presenting as a diastolic murmur, or with symptoms of heart failure. A detailed vascular examination is recommended to look for concomitant peripheral vascular diseases, carotid artery disease, or signs of peripheral embolization [1].

Echocardiography is the first-line examination for assessing the diameter of the ascending aorta and monitoring its progression over time [4].

Measurements should be performed using two-dimensional echocardiography, as M-mode is insufficient for this evaluation. Transesophageal echocardiography is useful for examining the anatomy of the aortic valve and ascending aorta, allowing differentiation between ascending aortic aneurysms, dissections, and intramural hematomas. However, visualization of the distal portion of the aneurysm may be limited.

Spiral thoracic computed tomography (CT) with contrast injection is often used for initial evaluation and follow-up of patients. It is complemented by three-dimensional reconstruction to improve measurement accuracy, identify the proximal and distal segments of the aneurysm, and differentiate between dissection, penetrating ulcer, or intramural hematoma.

In addition to cardiac anatomy, MRI can assess cardiac perfusion and myocardial function. However, MRI is generally more expensive and less accessible than thoracic CT.

In asymptomatic patients, medical treatment with beta-blockers and follow-up by echocardiography 1 or 2 times a year are recommended.

Beta-blockers have a negative inotropic and chronotropic effect. They also reduce the mean arterial pressure exerted on the aortic wall, especially during physical exertion. Their slowing effect on aortic root dilation is well-demonstrated, and the survival of patients with Marfan syndrome is improved [1,5]. Although this effect has not been established for ascending aortic aneurysms of other etiologies, such as bicuspid aortic valves, this treatment is recommended by most authors in all cases.

Surgical intervention is indicated urgently in cases of acute ascending aortic dissection or acute tamponade. Elective surgery for the ascending aorta is associated with a significantly lower mortality rate (5%) compared to emergency surgery.

A key factor in the decision to operate is the progression of the maximum diameter of the ectasia, requiring multiple imaging checks to detect a substantial increase in diameter over a specific period. Only very large aneurysms or those causing complications lead to immediate surgery. Given the natural history of ascending aortic aneurysms, surgery is deemed appropriate when the maximum diameter reaches 5 - 5.5 cm, depending on the etiology.

Symptoms due to aortic valve insufficiency or stenosis may be the first indication for surgery. During valve surgery, a decision must be made regarding moderate aortic dilation. About 25% of patients operated on for aortic insufficiency, with an aortic diameter >4 cm, later require aortic replacement surgery [1].

The recommendations you mentioned are indeed in line with the recent surgical approaches for managing ascending aortic aneurysms, as defined by the ESC (European Society of Cardiology in 2024. These guidelines emphasize individualized management based on the extent and characteristics of the aneurysms, as well as the condition of the aortic valve.

Isolated dilatation of the tubular ascending aorta: In this case, the standard technique involves the insertion of a supra-commissural tubular graft with a distal anastomosis just before the aortic arch, which effectively treats the aneurysm while preserving the surrounding structures.

Aneurysms extending proximally below the sino-tubular junction: When the aneurysm extends below the sino-tubular junction and involves the aortic sinuses, the surgical approach depends on the condition of the aortic annulus and valve:

- If the aortic valve cusps are supple, aortic valve-sparing techniques may be considered, including the David procedure (or aortic root repair surgery) or the Yacoub technique.
- If the valve is damaged or preservation is not feasible, composite replacement of the aortic root and valve using the Bentall procedure is recommended, where the aortic valve is replaced with a prosthesis and an anastomosis is performed to ensure the integrity of the blood flow.

These strategies aim to ensure durable repair while minimizing risk to the patient.

Illustrations

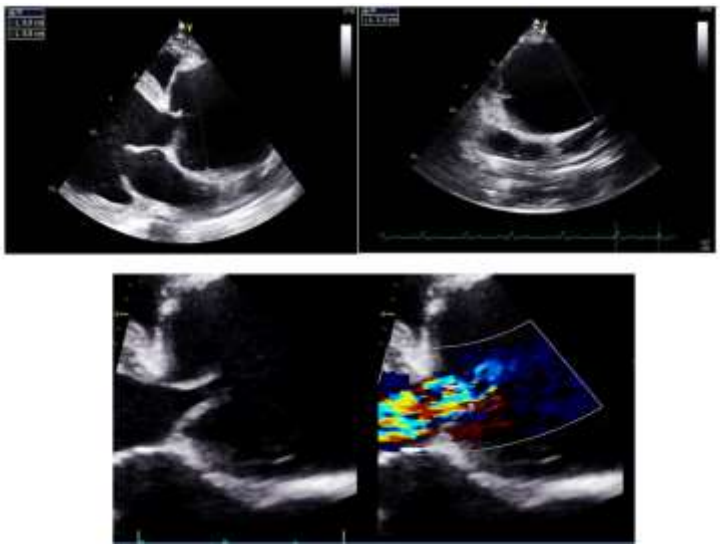

Fig.1: (TTE)***parasternal long-axis view*** *showing* ***ectasia of the initial part of the aorta*** *with* ***severe aortic insufficiency***

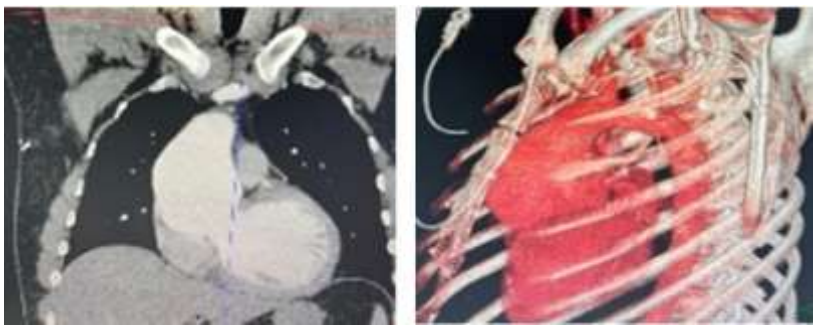

Fig.2: (aortic CT angiography) *significant dilation of the ascending aorta without dissection and without involvement of the aortic arch*

CONCLUSION

Aortic annuloectatic diseases, primarily including aneurysms of the aortic root and ascending aorta, represent significant clinical challenges due to their potential for severe complications, such as rupture or dissection. Management of these conditions relies on early diagnosis, often revealed by imaging studies like computed tomography (CT) and echocardiography, along with a thorough evaluation of surgical criteria. Current guidelines emphasize the importance of continuous monitoring and early surgical intervention for patients with critical diameters, particularly in the context of connective tissue diseases such as Marfan syndrome. A personalized approach, incorporating multidisciplinary assessment and rigorous follow-up, is essential to improve clinical outcomes and reduce the risks associated with these complex aortic pathologies.

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