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RESEARCH ARTICLE

ANNULOECTATIC AORTIC DISEASE OF MARFANOID ORIGIN: A CASE REPORT WITH LITERATURE REVIEW

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Abstract

Annuloectatic Disease is a group of pathologies with a similar phenotypic expression, combining aortic root aneurysm and dilation of the aortic ring. Echocardiography and spiral thoracic computed tomography are key examinations for assessing the diameter of the ascending aorta and its progression over time. Aortic root aneurysms can be idiopathic or associated with connective tissue disorders such as Marfan syndrome, Ehlers-Danlos syndrome, or bicuspid aortic valves. We present the case of a young patient from the cardiology department at HMIMV, who consulted for recurrent chest pain, and whose diagnostic evaluation revealed an ectasia of the aortic root.

Introduction:-

Annuloectatic aortic disease is a severe condition characterized by progressive dilatation of the aortic root, often leading to significant hemodynamic disturbances and an increased risk of aortic dissection or rupture. In patients with marfanoid features, this pathology is frequently linked to connective tissue disorders, most notably Marfan syndrome, which results from mutations in the FBN1 gene encoding fibrillin-1. The structural fragility of the aortic wall in these individuals predisposes them to life-threatening complications, underscoring the need for early detection and timely intervention. Advances in imaging modalities and surgical techniques have improved the management of this condition, yet challenges remain in optimizing long-term outcomes. This review explores the pathophysiology, clinical manifestations, diagnostic approaches, and current therapeutic strategies for annuloectatic aortic disease of marfanoid origin, highlighting the importance of a multidisciplinary approach in its management.

Case Description:

A 37-year-old man was referred to our Cardiology Center with a 2-month history of intermittent chest pain.
The patient has no cardiovascular disease in his clinical history.

The clinical examination identifies palpable thrills concomitant with a grade 5/6 diastolic murmur on the left sternal border, and also the presence of the Musset sign.

Transthoracic echocardiography (TTE) revealed an Aneurysm of the aortic root with severe aortic regurgitation.

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Aortic valve with three thin, showing central diastasis with significant dilation of the aortic root and obliteration of the Sino-tubular junction (LVOT: 31mm, VS: 73mm, TA: 75mm) with severe aortic regurgitation (ROA: 40mm², RV: 50ml, VC: 10mm, RJV: 0.4m/s).

The left ventricle (LV) is dilated, with non-hypertrophied walls, exhibiting hyperkinetic motion with systolic dysfunction at 44% SBP.

The aortic arch is not dilated (35mm), and the descending aorta measures 19mm without dilation**. (Figure A, B, C)**

The aortic CT (ACT) scan revealed an aneurysmal dilation primarily in segment I of the ascending aorta, without any signs of complications.**(Figure D, E,F)**

However, an etiological assessment was initiated, with laboratory tests showing no abnormalities. A cerebral and abdominal CT angiography, along with a Doppler ultrasound of the lower limbs, did not reveal any other aneurysmal locations.

Surgical intervention was indicated due to the significant size of the aneurysm and aortic regurgitation to prevent the risk of rupture or dissection. The surgical procedure involved a total replacement of the ascending aorta while preserving the native aortic valve, using the Tirone David technique, under cardiopulmonary bypass, with straightforward postoperative recovery.

Discussion:

Aortic aneurysms represent a major cardiovascular pathology and rank as the 13th leading cause of mortality in developed countries [1, 2]. The incidence of thoracic aortic aneurysms is estimated at approximately 4.5 cases per 100,000 individuals [1, 3]. Supravalvular aneurysms, although less prevalent than aortic root aneurysms, predominantly affect males, with an average age of onset between 59 and 69 years. Conversely, aortic root aneurysms occur at a younger age, typically between 30 and 50 years.

Aortic aneurysms are degenerative vascular disorders primarily driven by three pathological processes: proteolysis leading to extracellular matrix degradation, progressive loss of medial smooth muscle cells, and their replacement by areas of mucoid degeneration rich in vacuoles and sulfated polysaccharides. Aortic root aneurysms, categorized under annuloectatic aortic diseases, may arise idiopathically or be associated with hereditary connective tissue disorders such as Marfan syndrome, Ehlers-Danlos syndrome, or congenital bicuspid aortic valve anomalies [1, 3]. Approximately 20% of Marfan syndrome patients require surgical intervention for aortic root aneurysm repair. Furthermore, a significant association has been documented between bicuspid aortic valve disease and aortic dissection, with an approximately tenfold increased risk relative to the general population.

Most ascending aortic aneurysms remain clinically silent for prolonged periods and are often detected incidentally through imaging studies performed for unrelated cardiac conditions or during routine screening in individuals with a family history of connective tissue disorders. When symptoms develop, acute presentations—such as sudden, severe, tearing chest pain—are highly suggestive of impending rupture or dissection, occurring in approximately 75% of cases. Chronic pain, attributed to compressive effects on the sternum, is reported as the initial symptom in 25–75% of patients. Additional manifestations may result from external compression of adjacent structures, leading to symptoms such as cough, hemoptysis, dysphagia, dysphonia, superior vena cava syndrome, or, more rarely, aortocardiac or aortopulmonary fistulas [1].

Diagnostic Modalities

Clinical examination alone is often insufficient for diagnosing aortic aneurysms. However, auscultation may reveal a diastolic murmur indicative of aortic insufficiency or findings suggestive of heart failure. A thorough vascular assessment is essential to identify concomitant vascular involvement, including carotid artery disease and peripheral embolization [1].

Echocardiography serves as the primary imaging modality for evaluating aortic dimensions and monitoring disease progression [4]. Two-dimensional echocardiography is preferred over M-mode for precise measurements. Transesophageal echocardiography provides superior visualization of the aortic valve and ascending aorta,

facilitating differentiation between aneurysms, dissections, and intramural hematomas, though visualization of distal segments remains limited.

Contrast-enhanced spiral computed tomography (CT) is widely utilized for both initial diagnosis and longitudinal follow-up, offering high-resolution imaging and three-dimensional reconstructions to delineate aneurysm morphology and detect complications such as dissection or intramural hematomas. Magnetic resonance imaging (MRI) provides additional functional assessment of cardiac anatomy, perfusion, and myocardial contractility, though its use is limited by cost and accessibility constraints compared to CT.

Therapeutic Strategies

In asymptomatic patients, medical management with beta-blockers is recommended, alongside periodic echocardiographic surveillance every 6 to 12 months [1,5]. Beta-blockers exert negative inotropic and chronotropic effects, thereby reducing hemodynamic stress on the aortic wall and slowing aneurysm progression. In patients with Marfan syndrome, this pharmacological approach has demonstrated a significant survival benefit. While its efficacy in aneurysms of other etiologies, such as bicuspid aortic valve disease, remains less well established, beta-blocker therapy is widely advocated in all cases.

Surgical intervention is warranted in acute scenarios, including ascending aortic dissection or cardiac tamponade, where emergent repair is lifesaving. Elective surgical repair is associated with a significantly lower perioperative mortality rate (5%) compared to emergency interventions. The primary determinant for elective surgery is aneurysm size and its rate of expansion. Current guidelines recommend intervention when the maximal aortic diameter reaches 5–5.5 cm, depending on the underlying pathology. Additionally, aortic valve dysfunction, manifesting as significant regurgitation or stenosis, may serve as an indication for surgical correction.

Surgical Techniques

Recent guidelines from the European Society of Cardiology (ESC, 2024) emphasize a tailored surgical approach based on aneurysm extent and aortic valve pathology. The principal surgical strategies include:

- **Isolated dilatation of the ascending aorta**: Standard management consists of replacing the affected segment with a supracommissural tubular graft, ensuring anatomical preservation while restoring vascular integrity.
- **Aneurysms extending below the sinotubular junction**:
  - In cases where the aortic valve leaflets remain structurally viable, valve-sparing procedures such as the David or Yacoub techniques are preferred.
  - If valve preservation is unfeasible due to structural degeneration, composite aortic root replacement via the Bentall procedure is indicated. This approach involves implantation of a prosthetic valve with a vascular graft and coronary artery reimplantation.

These surgical techniques have significantly improved long-term outcomes, reducing postoperative complications and enhancing survival in patients with annuloectatic aortic disease of marfanoid origin.

Conclusion

Aortic annuloectatic diseases, encompassing aneurysms of the aortic root and ascending aorta, pose significant clinical challenges due to their high risk of life-threatening complications, including rupture and dissection. Effective management relies on early detection through advanced imaging modalities such as computed tomography (CT) and echocardiography, coupled with a meticulous assessment of surgical indications. Current guidelines highlight the necessity of ongoing surveillance and timely surgical intervention, particularly for patients with connective tissue disorders like Marfan syndrome, where disease progression is more aggressive. A tailored, multidisciplinary approach, combined with stringent long-term monitoring, is crucial for optimizing patient outcomes and mitigating the risks associated with these complex aortic conditions.

Illustrations

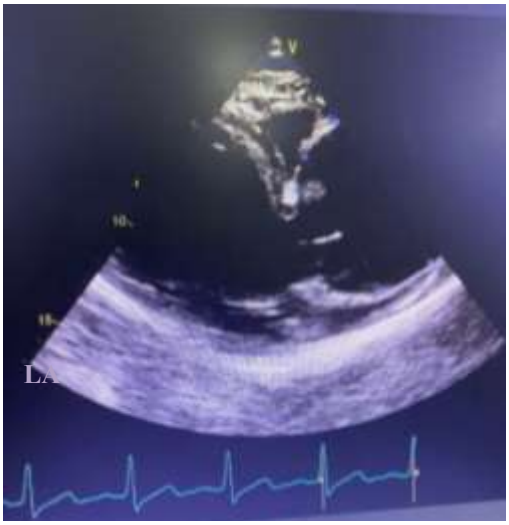

Figure A:- TTE parasternal long-axis view revealing an aneuvrysmal dilation of the aortic root.

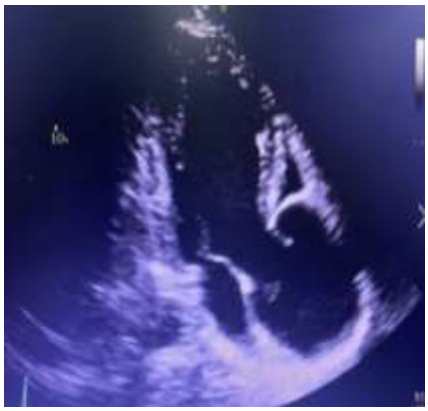

Figure B:- TTE apical three chambers view revealing an aneuvrysmal dilation of the aortic root.

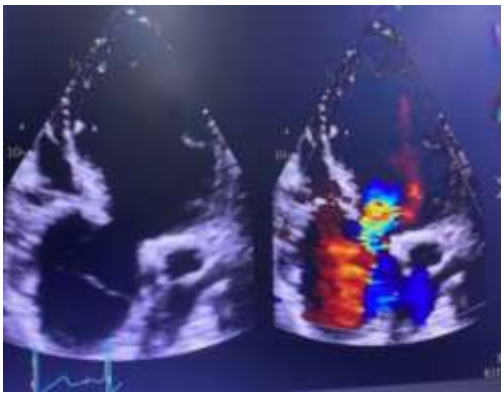

Figure C:- TTE apical five chambers view revealing an aneuvrysmal dilation of the aortic root with severe aortic regurgitation.

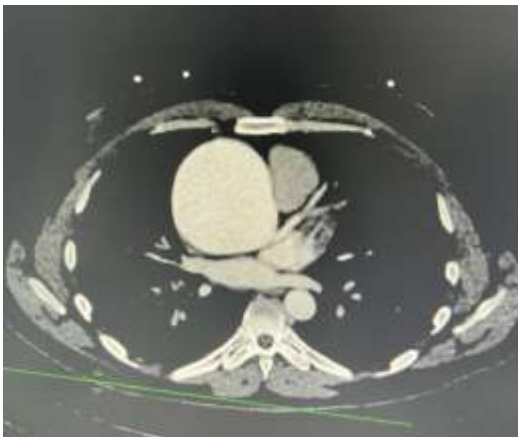

Figure D:- Axial view in mediastinal window on a contrast-enhanced thoracic CT scan, showing an aneurysmal dilation of the ascending aorta.

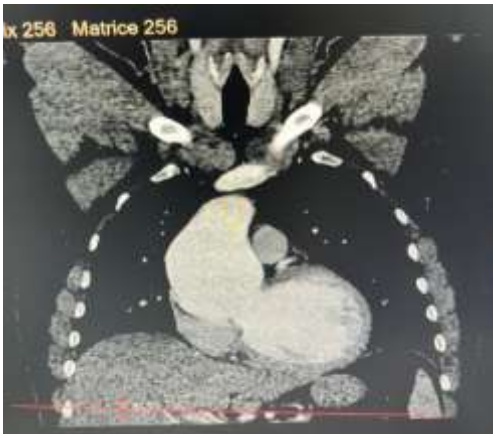

Figure E:- Coronal view on a contrast-enhanced CT scan, illustrating the aneurysmal dilation of the ascending aorta at its origin.

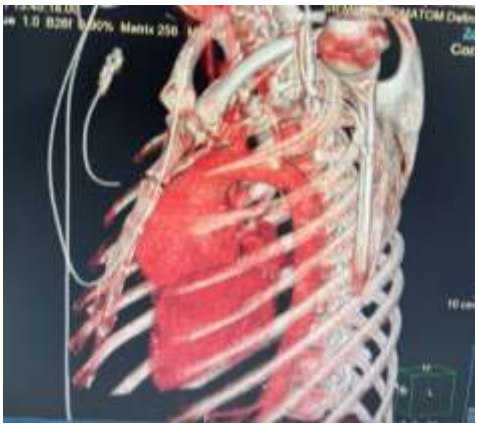

Figure F:- 3D reconstruction with a sagittal view, displaying the aneurysmal dilation of the thoracic aorta.

Consent:
The authors confirm that written consent for the submission and publication of this case, including images, has been obtained from the patient and his parents in line with the Committee on Publication Ethics (COPE) guidance.

Availability of Data and Materials:
Data sharing is not applicable to this article as no datasets were generated or analyzed during the current study.

Conflict of interest:
None declared.

Funding:
None declared.

Figure Legend: Tte And Act Findings
A: TTE parasternal long-axis view revealing an aneuvrysmal dilation of the aortic root.
B: TTE apical three chambers view revealing an aneuvrysmal dilation of the aortic root.
C: TTE apical five chambers view revealing an aneuvrysmal dilation of the aortic root with severe aortic regurgitation.
D:Axial view in mediastinal window on a contrast-enhanced thoracic CT scan, showing an aneurysmal dilation of the ascending aorta
E: Coronal view on a contrast-enhanced CT scan, illustrating the aneurysmal dilation of the ascending aorta at its origin.
F: 3D reconstruction with a sagittal view, displaying the aneurysmal dilation of the thoracic aorta.

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