CARDIOLOGIE TUNISIENNE

CASE REPORT

Giant Aortic Aneurysm Revealing Familial Aortopathy

Anévrisme aortique géant révélant une aortopathie familiale

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Summary

Aortic aneurysms represent a life-threatening vascular anomaly characterized by an abnormal enlargement of the aorta and carrying a high risk of complications such as rupture or dissection and sudden death if not detected and managed promptly. The etiology of aortic aneurysms encompasses degenerative and hereditary causes (syndromic and non-syndromic heritable thoracic aortic disease: HTAD). Imaging modalities, such as transthoracic echocardiography and computed tomography (CT) angiography, play a pivotal role in the diagnosis and management of aortic pathology. We report a case of a 27-year-old woman with a strong familial history of aortic dissection who was admitted for acute heart failure due to severe aortic regurgitation secondary to a giant aortic aneurysm. She successfully underwent a valve-sparing replacement of the aortic root, ascending aorta, aortic arch, and proximal descending aorta. The postoperative course was favorable. This case highlights the importance of early screening and close monitoring in individuals with a familial predisposition to aortic dissection and the critical role of timely surgical intervention

Keywords

aortic aneurysm, aortic regurgitation, valve-sparing surgery, screening, imaging, heritable thoracic aortic disease.

Résumé

Les anévrismes de l'aorte constituent une anomalie vasculaire potentiellement mortelle, caractérisée par une dilatation anormale de l'aorte et associée à un risque élevé de complications telles que la rupture, la dissection et la mort subite lorsqu'ils ne sont pas détectés et pris en charge à temps. Le bilan étiologique des anévrismes aortiques comprend des causes dégénératives et héréditaires, dont les maladies aortiques thoraciques héréditaires syndromiques et non syndromiques (HTAD). Les modalités d'imagerie, notamment l'échocardiographie transthoracique et l'angiographie par tomodensitométrie (TDM), jouent un rôle essentiel dans le diagnostic et la prise en charge des pathologies aortiques. Nous rapportons le cas d'une femme de 27 ans, présentant des antécédents familiaux de dissection aortique, admise pour une insuffisance cardiaque aiguë secondaire à une insuffisance aortique sévère due à un anévrisme aortique géant. La patiente a bénéficié avec succès d'un remplacement de la racine aortique, de l'aorte ascendante, de la crosse aortique et de la portion proximale de l'aorte descendante avec préservation valvulaire. L'évolution postopératoire a été favorable. Ce cas souligne l'importance du dépistage précoce et d'une surveillance étroite chez les individus présentant une prédisposition familiale à la dissection aortique, ainsi que le rôle crucial d'une intervention chirurgicale réalisée en temps opportun.

Mots-clés

anévrisme de l'aorte ; insuffisance aortique; chirurgie valvulo-épargnante ; dépistage; imagerie; maladie aortique thoracique héréditaire

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INTRODUCTION

Thoracic aortic aneurysms represent a progressive and potentially fatal pathology that is often asymptomatic until acute complications such as dissection or rupture occur. The risk of such events is strongly correlated with aortic diameter, with a marked increase beyond 5.5–6.0 cm. It is estimated that aortic aneurysms are responsible for approximately 150,000 to 200,000 deaths annually around the globe (1,2). Aneurysms involving the ascending aorta and aortic root are frequently linked to underlying genetic predispositions, including both syndromic (e.g., Marfan syndrome) and non-syndromic forms of heritable thoracic aortic disease (HTAD) . In contrast, aneurysms of the descending thoracic aorta are more often degenerative in origin and associated with advanced age and atherosclerosis (3). Diagnostic workup of thoracic aortic aneurysms relies on multimodal imaging, including chest radiography, transthoracic and transesophageal echocardiography, contrast-enhanced computed tomography (CT), and magnetic resonance angiography. Among these, contrast-enhanced CT remains the gold standard for detailed anatomical evaluation of aneurysm size, extent, and involvement of adjacent structures. While conservative (non-surgical) management is generally associated with poor long-term outcomes, especially in large or symptomatic aneurysms, surgical repair remains the main curative option. However, operative risks increase significantly when surgery involves concomitant aortic valve replacement or total arch reconstruction (4).

CASE REPORT

Thi We present the case of a 27-year-old woman referred for acute heart failure due to severe aortic regurgitation. Her medical history was unremarkable; however, her family history was notable for multiple cases of aortic aneurysm and dissection: her father, paternal aunt, and a first cousin underwent surgical repair for thoracic aortic aneurysm, while a brother and another cousin died suddenly due to aortic rupture.

On admission, the patient exhibited signs of left-sided heart failure. There were no phenotypic stigmata suggestive of a syndromic connective tissue disorder.

Transthoracic echocardiography (TTE) revealed a tricuspid aortic valve with preserved leaflet morphology but significant annulo-aortic ectasia leading to severe central aortic regurgitation. The sinus of Valsalva measured 38 mm, the sino-tubular junction 49 mm, and the ascending aorta 66 mm. Left ventricular dilation with a reduced ejection fraction of 40% was observed (Figures 1 and 2).

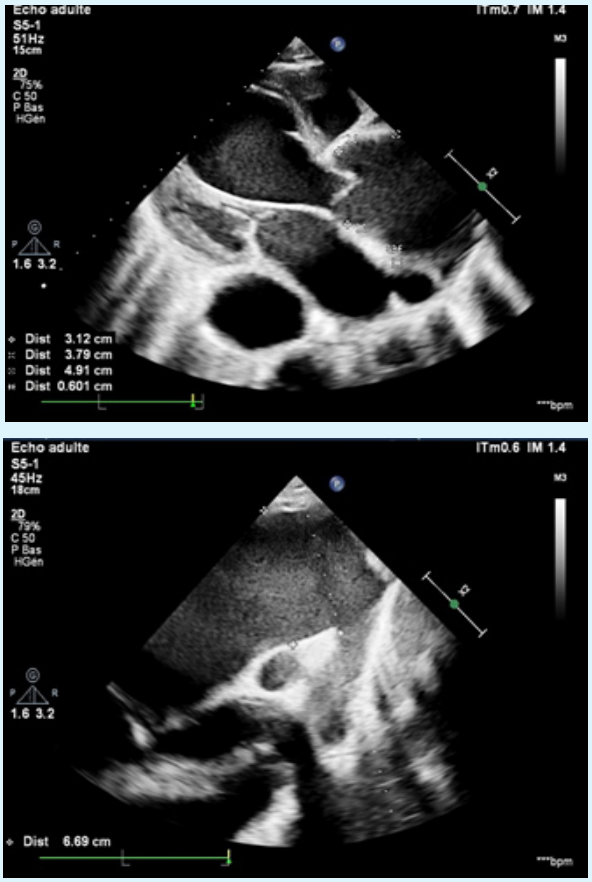

Figure 1. transthoracic echocardiography A: Para sternal long-axis view B: Supra sternal view: Giant aortic aneurysm extending from the annulus to the origin of the left subclavian artery

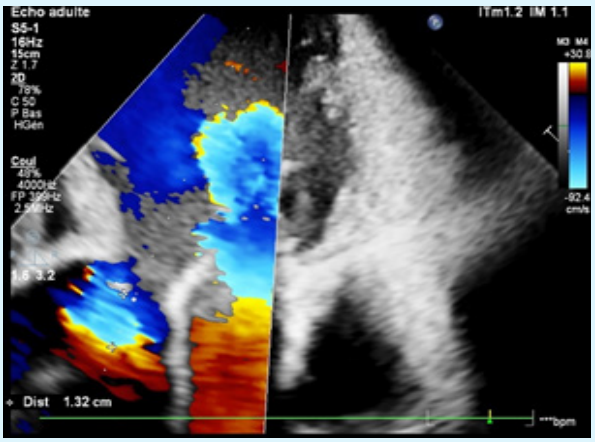

Figure 2. Severe aortic regurgitation secondary to the aneurysm of the proximal aorta.

Contrast-enhanced computed tomography (CT) angiography demonstrated a markedly aneurysmal ascending aorta (maximum diameter: 82 mm) extending to the transverse aortic arch (up to 73 mm), without evidence of dissection. The isthmus was of normal caliber. A fusiform aneurysm of the descending thoracic aorta was also identified (maximum diameter: 50 mm), along with congenital agenesis of the left common iliac artery (Figure 3).

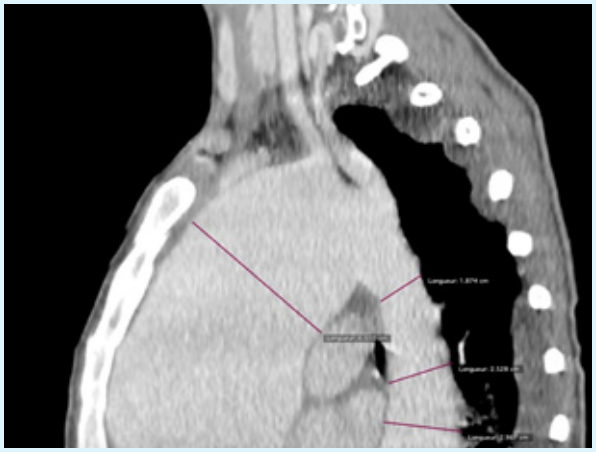

Figure 3. Contrast-enhanced computed tomography (CT) angiography demonstrated an aneurysmal ascending aorta (maximum diameter: 82 mm) extending to the transverse aortic arch

During hospitalization, the patient developed cardiogenic shock requiring inotropic support. After stabilization, she underwent surgical intervention under cardiopulmonary bypass (CPB) via median sternotomy. CPB was initiated between the right atrium and the axillary artery, providing antegrade cerebral perfusion. Under moderate hypothermia (25°C) and circulatory arrest, the ascending, arch, and proximal descending aorta were replaced with a Dacron graft with reimplantation of the supra-aortic trunks as a single island. The native aortic valve was preserved (Figure 4).

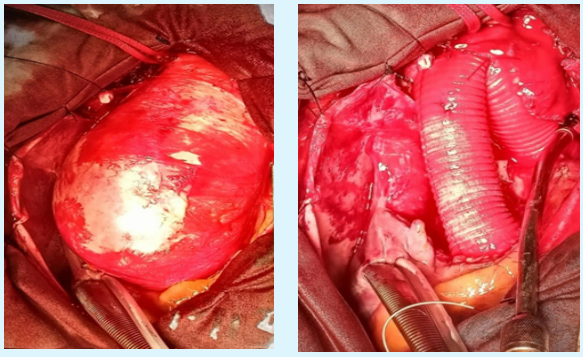

Figure 4. Surgical replacement of the ascending, arch, and proximal descending aorta with a Dacron graft.

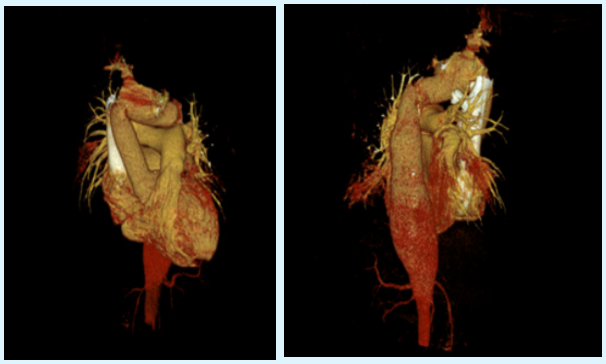

Figure 5. postoperative CT scan showing the Dacron graft of the ascending aorta and transverse aortic arch

DISCUSSION

Thoracic aortic aneurysms (TAAs) are more prevalent in men than in women (ratio 4:1); however, the growth rate is greater in women (0.96 ± 1.00 mm per year) than in men (0.45 ± 0.58 mm per year), and the average age at diagnosis is around 65 years (5). In our case, the patient was a 26-year-old female, highlighting an uncommon presentation at a much younger age compared to typical demographics.

Anatomically, approximately 60% of TAAs involve the aortic root or ascending aorta, 10% affect the aortic arch, 40% are located in the descending thoracic aorta, and 10% occur in the thoracoabdominal segment. Notably, aneurysmal disease may simultaneously involve multiple aortic segments (5). In our case, aneurysms include the ascending aorta extending to the transverse aortic arch and descending aorta.

The etiology of TAA, particularly affecting the root and ascending aorta, includes several underlying causes: heritable thoracic aortic disease (HTAD), bicuspid aortic valve (BAV), sporadic TAA, and atherosclerosis (6).

Inherited aortic diseases are typically classified into two main groups depending on the clinical presentation: syndromic heritable thoracic aortic disease (sHTAD) and non-syndromic heritable thoracic aortic disease (nsHTAD). sHTAD includes genetic syndromes such as Marfan syndrome and Loeys-Dietz syndrome, characterized by systemic manifestations. In contrast, nsHTAD is defined by familial occurrences of aortic dissection and dilatation at an early age without associated systemic or syndromic features (7).

The genetic basis of non-syndromic hereditary thoracic aortic disease (nsHTAD) is diverse, involving mutations in key genes such as ACTA2, MYH11, MYLK, LOX, PRKG1, FOXE3, TGFBR1, TGFBR2, SMAD3, TGFB2, COL3A1, and FBN1. Despite this genetic heterogeneity, only approximately 20% of nsHTAD cases have

identifiable pathogenic mutations, leaving the majority of patients without a definitive genetic diagnosis. (7-9).

Approximately 20% of individuals with thoracic aortic aneurysms or dissections have a family history. Therefore, genetic screening for hereditary thoracic aortic disease (HTAD) can play a crucial role in identifying causal genetic mutations, recognizing asymptomatic family members at risk, and determining the best timing for preventive surgical interventions in those diagnosed with genetic aortopathy (10).

Thoracic aortic aneurysms are often asymptomatic but can lead to life-threatening complications like aortic dissection or rupture if not detected in time. In larger aneurysms, clinical symptoms may be present due to compression or erosion of surrounding structures and organs. When aneurysms occur in the aortic root or ascending aorta, they can cause secondary aortic regurgitation, which may be revealed by the presence of a diastolic murmur during physical exam.

Multimodality imaging plays a central role in evaluating thoracic aortic aneurysms. Several imaging techniques are used for a comprehensive assessment, including computed tomography (CT), magnetic resonance imaging (MRI), transthoracic echocardiography (TTE), and transesophageal echocardiography (TEE). Echocardiography remains the first-line tool, as it allows detailed evaluation of the aortic root and the proximal ascending aorta. Additionally, it provides valuable information on valve anatomy, enables quantification of aortic regurgitation, and helps identify its underlying mechanisms, thus aiding in the selection of appropriate surgical strategies (11). The decision to proceed with surgery should ideally be based on measurements from computed tomography (CCT), which assess the maximum diameter (12).

When the aortic diameter reaches 57.5 mm, the annual incidence rates for rupture, dissection, and death are 3.6%, 3.7%, and 10.8%, respectively (13).

Conservative management of ascending aortic aneurysms (AscAA) is associated with poor long-term survival. Surgical intervention remains the main definitive treatment, although it is associated with significant procedural risk.

A meta-analysis reviewing 3794 patients showed no significant differences in early outcomes such as mortality and myocardial infarction between two surgical approaches: aortic valve-sparing and composite valve-grafting (bioprosthetic and mechanical) procedures. However, long-term results favored aortic valve-sparing, demonstrating a lower rate of late mortality and a substantially reduced risk of thromboembolic events and bleeding. Importantly, the long-term durability of the two procedures was found to be equivalent (14).

CONCLUSION

T This case highlights the critical importance of early screening for aortic aneurysms in non-syndromic patients, particularly those with a significant family history of aortic dissection. Timely intervention proved vital for our patient, as it addressed severe aortic regurgitation and mitigated the risk of life-threatening complications. Moreover, the involvement of an expert surgical center specializing in valve-sparing techniques was instrumental in optimizing the surgical outcome. Overall, optimal management of HTAD involves lifelong multidisciplinary care, guided by genetic information, along with regular imaging surveillance and appropriate lifestyle considerations to improve clinical outcomes

REFERENCES

1. Goldfinger JZ, Halperin JL, Marin ML, Stewart AS, Eagle KA, Fuster V. Thoracic Aortic Aneurysm and Dissection. J Am Coll Cardiol. oct 2014;64(16):1725-39.
2. Cho MJ, Lee MR, Park JG. Aortic aneurysms: current pathogenesis and therapeutic targets. Exp Mol Med. déc 2023;55(12):2519-30.
3. Vapnik JS, Kim JB, Isselbacher EM, Ghoshhajra BB, Cheng Y, Sundt TM, et al. Characteristics and Outcomes of Ascending Versus Descending Thoracic Aortic Aneurysms. Am J Cardiol. 15 mai 2016;117(10):1683-90.
4. Isselbacher EM, Preventza O, Hamilton Black J, Augoustides JG, Beck AW, Bolen MA, et al. 2022 ACC/AHA Guideline for the Diagnosis and Management of Aortic Disease: A Report of the American Heart Association/American College of Cardiology Joint Committee on Clinical Practice Guidelines. Circulation. 13 déc 2022 [cité 23 avr 2025];146(24).
5. Isselbacher EM. Thoracic and Abdominal Aortic Aneurysms. Circulation. 15 févr 2005;111(6):816-28.
6. Coady MA, Rizzo JA, Goldstein LJ, Elefteriades JA. NATURAL HISTORY, PATHOGENESIS, AND ETIOLOGY OF THORACIC AORTIC ANEURYSMS AND DISSECTIONS. Cardiol Clin. 1 nov 1999;17(4):615-35.
7. Fletcher AJ, Syed MBJ, Aitman TJ, Newby DE, Walker NL. Inherited Thoracic Aortic Disease: New Insights and Translational Targets. Circulation. 12 mai 2020;141(19):1570-87.
8. Weerakkody R, Ross D, Parry DA, Ziganshin B, Vandrovcova J, Gampawar P, et al. Targeted genetic analysis in a large cohort of familial and sporadic cases of aneurysm or dissection of the thoracic aorta. Genet Med. nov 2018;20(11):1414-22.
9. Wolford BN, Hornsby WE, Guo D, Zhou W, Lin M, Farhat L, et al. Clinical Implications of Identifying Pathogenic Variants

in Individuals With Thoracic Aortic Dissection. Circ Genomic Precis Med. juin 2019;12(6):e002476.

10. Acharya M, Maselli D, Mariscalco G. Genetic screening in heritable thoracic aortic disease—rationale, potentials and pitfalls. Indian J Thorac Cardiovasc Surg. avr 2022;38(Suppl 1):24-35.
11. Evangelista A, Flachskampf FA, Erbel R, Antonini-Canterin F, Vlachopoulos C, Rocchi G, et al. Echocardiography in aortic diseases: EAE recommendations for clinical practice. Eur J Echocardiogr. 1 sept 2010;11(8):645-58.
12. Vahanian A, Beyersdorf F, Praz F, Milojevic M, Baldus S, Bauersachs J, et al. 2021 ESC/EACTS Guidelines for the management of valvular heart disease: Developed by the Task Force for the management of valvular heart disease of the European Society of Cardiology (ESC) and the European Association for Cardio-Thoracic Surgery (EACTS). Rev Esp Cardiol Engl Ed. juin 2022;75(6):524.
13. Coady MA, Rizzo JA, Hammond GL, Mandapati D, Darr U, Kopf GS, et al. What is the appropriate size criterion for resection of thoracic aortic aneurysms? J Thorac Cardiovasc Surg. mars 1997;113(3):476-91.
14. Elbatarny M, Tam DY, Edelman JJ, Rocha RV, Chu MWA, Peterson MD, et al. Valve-Sparing Root Replacement Versus Composite Valve Grafting in Aortic Root Dilation: A Meta-Analysis. Ann Thorac Surg. 1 juill 2020;110(1):296-306.