Unicuspid Aortic Stenosis: Case Report

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Introduction

Unicuspid aortic valve (UAV) is an uncommon congenital pathology, first described by Edwards in 1958.[1] It is a rare malformation (especially when compared to other aortic valve anomalies), with an estimated incidence of 0.02% of adults undergoing echocardiography and approximately 4% to 5% of patients undergoing surgery for aortic stenosis. Echocardiography is the first-choice method for diagnosis and definition of hemodynamic repercussions.[2] In this study, we report the case of a patient with symptomatic aortic stenosis and describe her diagnosis and therapeutic management.

Case presentation

A 43-year-old female patient was admitted to our service, reporting progressive dyspnea (upon minimal effort, at admission) and mild, stabbing chest pain, which had begun approximately 3 months prior. She had hypertension and was under regular follow-up. Physical examination revealed a systolic murmur, predominantly in the aortic and accessory aortic foci, 4+/6+, radiating to the suprasternal notch. A transthoracic echocardiogram was requested, which revealed the following: concentric left ventricular hypertrophy; preserved left ventricular systolic function (with an estimated ejection fraction of 63%); grade II diastolic dysfunction; aortic valve with transvalvular peak systolic velocity of 4.38 m/s; maximum transvalvular gradient of 77 mmHg and mean of 51 mmHg (Figure 1); Doppler velocity index of 0.24; valve area of 0.76 cm^2 estimated by the continuity equation; mild reflux; and mitral valve with normal appearance and mild regurgitation. The morphological aspect of the aortic valve was difficult to characterize on transthoracic echocardiography, and an associated transesophageal assessment was performed, which revealed a unicuspid, unicommissural aortic valve (Figure 2), whose aspect was corroborated by 3-dimensional assessment (Figure 3), without other significant associated valvular lesions. Thoracic and abdominal aortic angiotomography was performed, showing normal calibers and regular contours. Coronary angiotomography did not show any obstructive lesions or anomalous paths. Surgical treatment was chosen, and the patient underwent valve replacement with a metal prosthesis (following discussion of the case with the patient), with good postoperative recovery and hospital discharge with outpatient follow-up.

Discussion

Congenital aortic valve malformation is a common cause of aortic valve disease, and it requires surgical treatment. Bicuspid aortic valve is the most common form, accounting for 90% of congenital malformations of the aortic valve. UAV is a relatively significant cause of stenosis in young people. Although it is significantly predominant in men, it has not been proven to be hereditary, and no correlations have been established with any specific genetic anomaly. Before the advent of improved echocardiographic techniques, its diagnosis was mainly based on intraoperative specimens or autopsy findings.[2,3]

According to the number of commissures, UAV can be classified as acommissural (with 3 underdeveloped, congenitally fused commissures) or unicommissural (2 underdeveloped commissures and 1 normal commissure, usually located between the left coronary and non-coronary cusps).[4] The acommissural subtype is more common in children, due to the severely and prematurely reduced opening area. In adults, the unicommissural form is the most prevalent, commonly presenting with dyspnea, angina, or syncope, resulting from valvular stenosis, typically around the third to fifth decade of life. Valvular regurgitation may be associated in varying degrees, but isolated presentation is rare.[5] Although the association is rarely described in the literature, UAV may predispose to infective endocarditis.[6]

Echocardiography is the method of choice for diagnosing UAV. The initial transthoracic approach

Keywords

Aortic Valve Stenosis; Echocardiography; Heart Valve Diseases.

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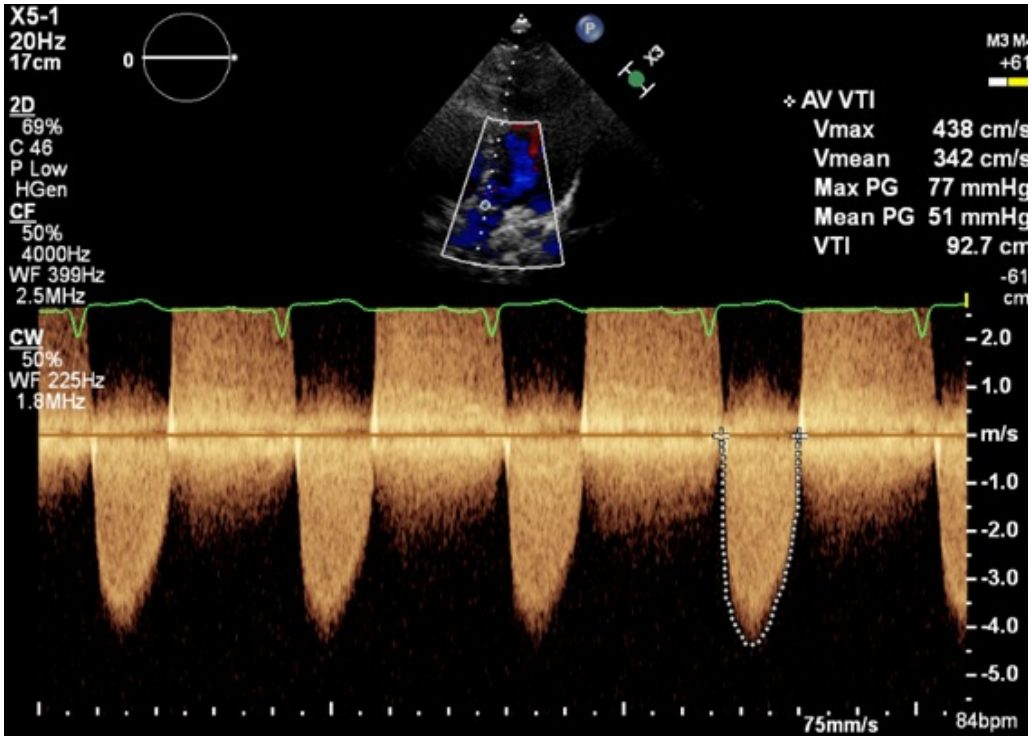

Figura 1 – *Análise de fluxo transvalvar aórtico, ao Doppler contínuo, evidenciando altas velocidade e gradientes*

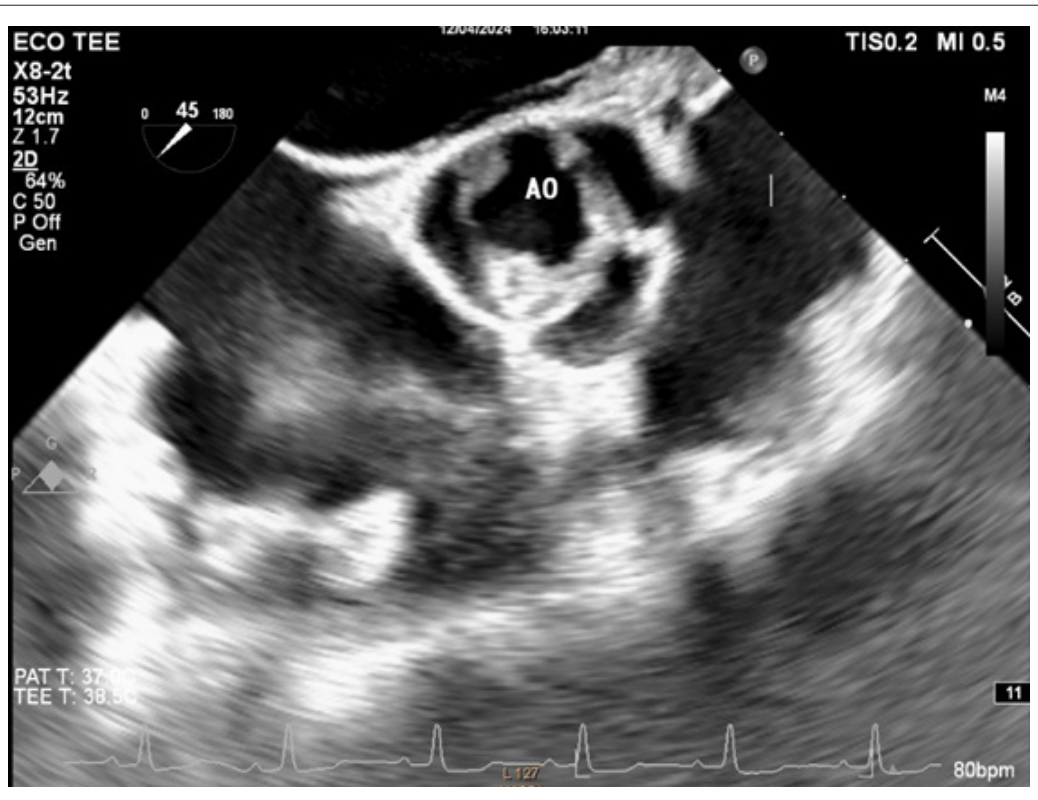

Figura 2 – *Visão transesofágica de VAU, com abertura limitada.*

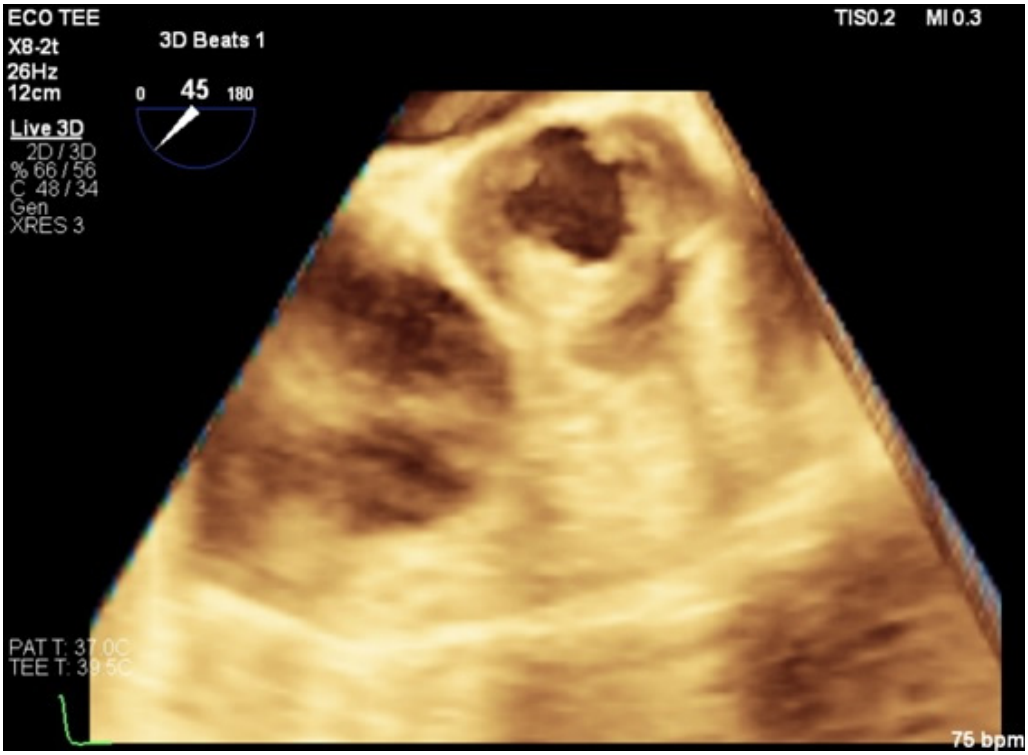

Figura 3 – *Visualização tridimensional de VAU.*

may be hindered by limited acoustic windows, in addition to an eventual high degree of diffuse valve calcification and reduced cusp mobility. In contrast, transesophageal examination has significantly higher diagnostic sensitivity (75%) and specificity (86%). The following findings have been commonly associated with diagnosis of the condition: in the short-axis view, a posteriorly located orifice with 1 or 2 anteriorly visible raphes or a lone commissure located between the left and non-coronary cusps (both indicative of UAV); in the long-axis view, the coaptation line of the cusps located eccentrically towards the interventricular septum may also be suggestive. Surgical diagnosis has been considered the gold standard.[3] Other complementary methods, such as tomography, play a role in the assessment of the disease, given the need to evaluate other anomalies that are commonly present (for example, aortic or coronary anomalies), as well as eventual cases where echocardiography is limited in defining preoperative valve anatomy in greater detail.[7]

The treatment of UAV can be challenging, given that the surgical modality chosen must respect the generally long life expectancy of patients who are usually young when diagnosed, as well as their quality of life and functionality. The choice of the best strategy should be thoroughly discussed with the patient and adapted to their individual needs, also taking the expertise of the treatment center into account. The surgical technical principles are divided between sparing the native valve and repairing it or replacing it with a prosthesis (mechanical, biological, or pulmonary autografts, also known as the Ross procedure).[4] In patients with high surgical risk and favorable anatomy, transcatheter aortic valve implantation may be considered; however, there are fewer reports in the literature, given the frequent technical limitations to this method, for instance, the frequent presence of aortopathy and eccentric valve opening, which can make valve crossing and device delivery difficult.[8]

Conclusion

Even though it is rare, UAV is a disease that should be part of the differential diagnosis of aortic valve diseases in adults, especially when more common causes have not been clearly associated with the case. In these cases, it is up to the assisting team to use multimodality in cardiovascular imaging, and echocardiography is the first-choice exam in the search for a precise diagnosis.

Author Contributions

Conception and design of the research and writing of the manuscript: Sousa WGS; acquisition of data: Sousa WGS, Domingues JL, Lemos AET, Araújo ALJ, Damasceno AT, Evangelista NL; critical revision of the manuscript for intellectual content: Domingues JL, Lemos AET, Araújo ALJ, Damasceno AT, Evangelista NL.

Potential Conflict of Interest

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Study Association

This study is not associated with any thesis or dissertation work.

Ethics Approval and Consent to Participate

This article does not contain any studies with human participants or animals performed by any of the authors.

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