

Ebstein's Anomaly Associated with Left Heart Malformations: Presentation of Two Cases and Review of the Literature

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Abstract

Introduction: Ebstein's anomaly is a rare congenital cardiac malformation. This anomaly is seen as part of a generalized disturbance in the development of the right ventricle and, in some cases, of the left ventricle.

Case presentation: We present two adult women with this anomaly; both with mitral valve regurgitation, one of them with prolapse of the anterior leaflet and the other one with cleft of the anterior mitral leaflet, and the last one have also bicuspid aortic valve, aortic coarctation, persistence of ductus arteriosus and ostium secundum atrial septal defect.

Conclusion: The non-invasive imaging techniques such as echocardiography and cardiac magnetic resonance imaging play a very important role in the diagnosis, prognosis and treatment of these patients. An understanding of the morphological and functional details of Ebstein's anomaly is the most important point for the correct interpretation of the noninvasive images and adequate clinical judgment. Finally, it is mandatory to assess the left cavities of the heart in all patients with Ebsteins anomaly, in order to establish a complete evaluation.

Keywords: Ebstein's anomaly; Echocardiography; Mitral regurgitation; Bicuspid aortic valve; Aortic coarctation

Abbreviations: EA: Ebstein's Anomaly; ECG: Electrocardiogram; MRI: Magnetic Resonance Imaging; RA-Right atrium, RV: Right Ventricle; LA: Left Atrium; LV: Left Ventricle; TV: Tricuspid Valve; MV: Mitral Valve; AL: Anterior Leaflet; PL: Posterior Leaflet; ASD: Atrial Septal Defect; ARV: Atrialized Portion of the Right Ventricle; FRV: Functional Portion of the Right Ventricle; IRV: Infundibulum of the Right Ventricle; RB: Right Branch of the Pulmonary Artery; LB: Left Branch of the Pulmonary Artery

Introduction

Ebstein's anomaly (EA) is a rare congenital cardiac malformation. Its features are tethering of the leaflets to the ventricular wall, redundancy and dysplasia of the tricuspid valve leaflets, which determine the displacement of the functional tricuspid orifice toward the trabecular portion and outflow tract of the right ventricle [1]. The clinical manifestations of this cardiac malformation depend on the degree of tricuspid valve malformation. EA is regarded as a disease confined to the right side of the heart.

Objective: To describe two cases of adult women with Ebstein's anomaly associated with left heart malformations.

Case presentation 1

A 74-year-old female patient who routinely underwent cardiovascular evaluation for programmed inguinal hernia surgery, which was performed without any complications. She entered to a study protocol for the heart murmur found in the pre surgical assessment. She remained asymptomatic until November 2013, posteriorly she began with dyspnea of medium efforts and palpitations, during the physical exploration an holosystolic murmur grade III/IV was heard at the left lower parasternal border, which increased during inspiration and a systolic murmur with irradiation at the axilla grade II/IV and a rhythm of four times, edema of lower limbs and ascites. She received diuretics, inotropic, anticoagulant and oxygen treatment. The electrocardiogram (ECG) demonstrated atrial fibrillation, incomplete right bundle branch

block and volume overload of the right ventricle. The chest x-ray showed a severe cardiomegaly due to an elongated and enlarged right atrium, the heart has a "box shape". The 2D and 3D echocardiograms showed mild displacement and marked dysplasia of the free portion of the septal leaflet, tethering of the anterior and posterior leaflets, without coaptation, which cause a severe laminar tricuspid regurgitation. Also, prolapse of the anterior mitral leaflet was observed with moderate to severe mitral regurgitation, (Figure 1). The anatomico-echocardiographic correlation with the corresponding image was very good (Figure 2). Currently she is NYHA functional class III, continuing with same medical treatment and being monitored in the cardiology out-patient-clinic.

Case presentation 2

Female of 29 years-old with history of cardiac murmur from childhood, she remained asymptomatic until September 2014 that began with dyspnea of great efforts and palpitations once a week lasting 5 to 10 minutes. She attended to the emergency room and during chest auscultation a holosystolic tricuspid murmur grade IV/IV, which increased with the Rivero-Carvalho maneuver and holosystolic mitral murmur grade III/IV radiated to the axilla and third and fourth sounds were heard. Also hepatomegaly with hepatalgia was detected. The ECG demonstrated a complete right bundle branch block and a first-degree atrioventricular block. The chest X-ray showed a severe cardiomegaly (cardiothoracic index of 0.58). The 2D and 3D echocardiograms showed

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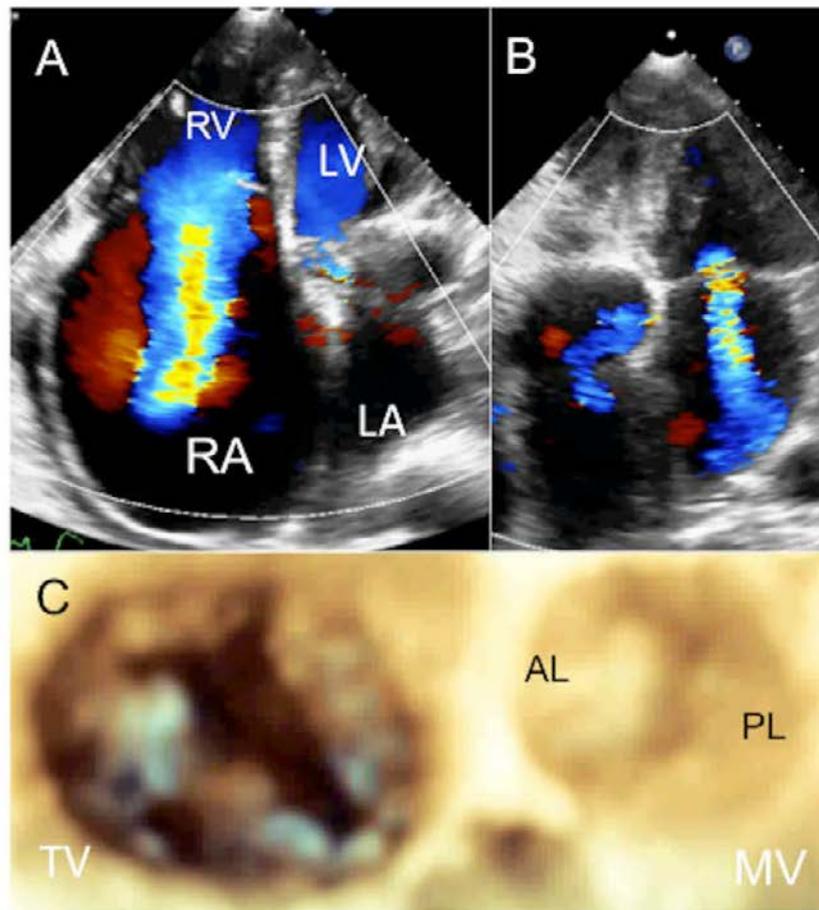


Figure 1: Bidimensional and color Doppler four chamber view showing a displacement of the septal leaflet of the tricuspid valve with severe laminar tricuspid regurgitation (A) and moderate to severe mitral regurgitation (B). A tridimensional atrial view of the atrioventricular valves is observed with prolapse of the anterior leaflet of the mitral valve and lack of coaptation of the tricuspid leaflets (C).

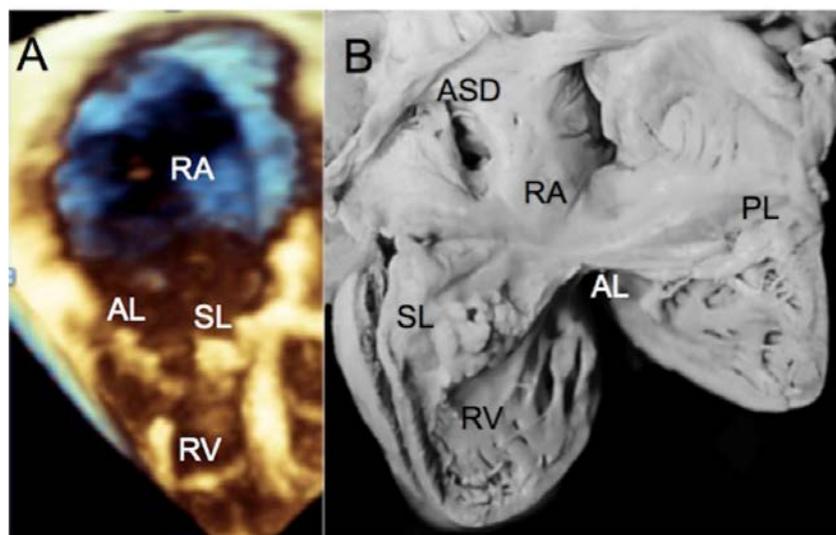


Figure 2: The 4-chamber 3D echocardiographic image shows mild displacement of the septal leaflet of the tricuspid valve and dysplasia of its free portion (A). In the anatomic specimen Internal view of the right chambers of a heart with Ebstein's anomaly a mild displacement of the septal leaflet and dysplasia of the free portion of the leaflet is evident. The atrialized portion of the right ventricle is small (B).

displacement of the septal leaflet and restrictive movement of the anterior leaflet of the tricuspid valve, without coaptation, which causes a severe tricuspid regurgitation (Figure 3). Also, cleft of the anterior mitral leaflet with moderate mitral regurgitation (Figure 4), bicuspid aortic valve without valvular lesions, postductal aortic coarctation, small patent ductus arteriosus and ostium secundum atrial septal defect were detected (Figure 5). These findings were corroborated in the cardiac magnetic resonance imaging (MRI), Figure 6. Now, she is NYHA functional class I with medical treatment (diuretic and antiplatelet agent).

Comment

Ebstein's anomaly is seen as part of a generalized disturbance in the development of the right ventricle and, in some cases, of the left ventricle. The left ventricle may be small in patients with Ebstein anomaly owing to severe tricuspid valve regurgitation, that progresses to left ventricular dysfunction in late stages, as a result of loss of ventricular-ventricular interaction [2]. Gerlis et al. described Ebstein malformation of both right and left atrioventricular valves, and accessory orifices in the valvular leaflets. In this study, the aortic valve was atretic [3]. Mitral prolapse have been reported [4-6], but in very rare cases involvement of the aortic valve [7,8] and aorta as we observed

in one of our patients. Cases with mild degree of leaflet tethering can remain asymptomatic until adulthood [9]. Leaflet tethering, together with dilatation of the tricuspid valve ring, constitute the anatomic cause of the tricuspid regurgitation observed in this condition.

Echocardiography especially three-dimensional is the non-invasive method of choice in its diagnosis, it allows to evaluate the degree of leaflet tethering, the characteristics of the leaflets and the subvalvular apparatus. The degree of regurgitation and/or tricuspid stenosis, the morphological and functional alterations in the atrialized right ventricle and associated anomalies in the left side of the heart, all of which aid in planning the type of treatment.

The comparison of anatomic specimens with corresponding echocardiographic images of different degrees of septal valve displacement has the potential of enhancing the echocardiographer's understanding of this anomaly. This comprehension can contribute to more precise diagnoses and lead to early and appropriate treatment of patients with a wide spectrum of manifestations of this condition. In some cases the MRI plays a very important role, especially in the assessment of extracardiac lesions such as the aortic coarctation and patent ductus arteriosus as we observed in one of our cases.

In our previous series we found alterations of the mitral valve

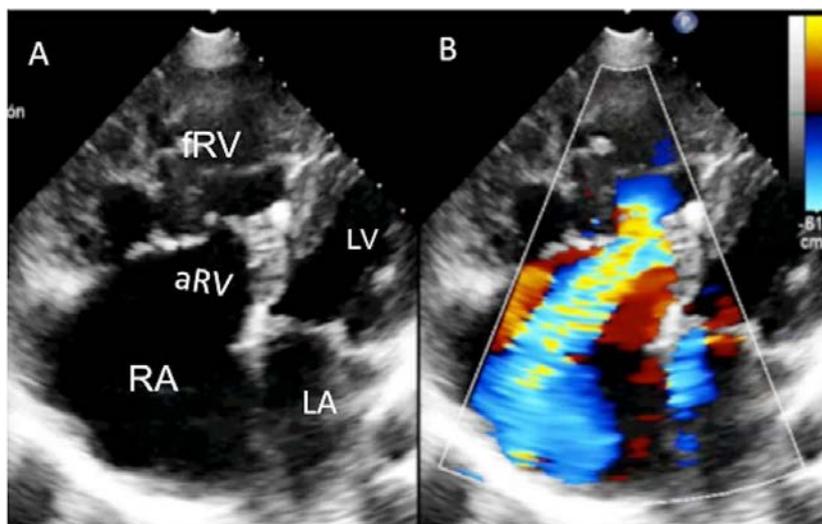


Figure 3: In bidimensional four chamber view the tricuspid valves are thickened and displacement of the septal leaflet, the atrialized and the functional portion of the right ventricle are visualized (A). With color Doppler a severe tricuspid regurgitation is observed (B).

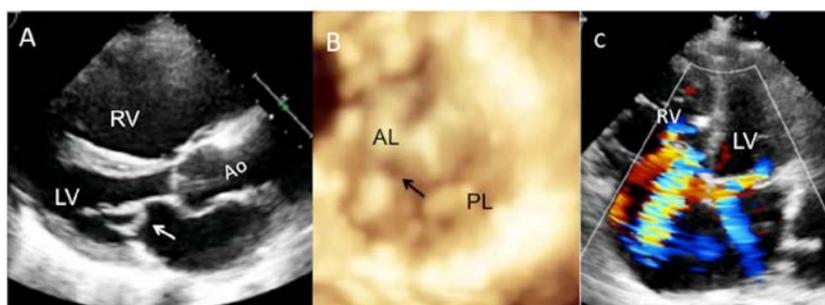


Figure 4: Bidimensional long axis view (A), tridimensional atrial view of the mitral valve (B) and color Doppler (C) images showing a cleft of the anterior leaflet (white and black arrows) of the mitral valve with moderate mitral regurgitation. Also a severe tricuspid regurgitation is observed.

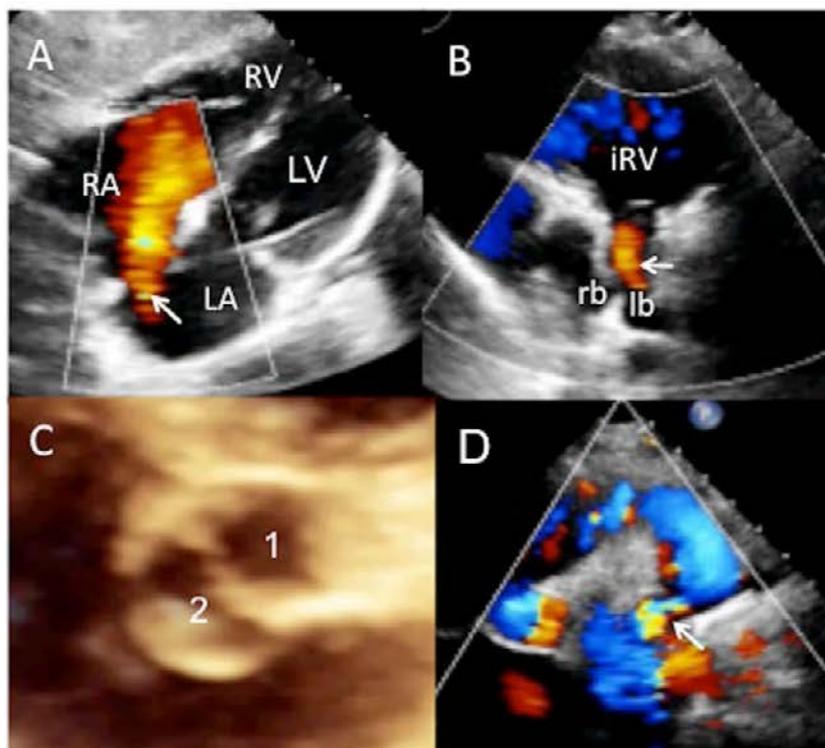


Figure 5: Color Doppler subcostal, parasternal, suprasternal and tridimensional images showing atrial septal defect (A), dilatation of the infundibulum and persistence of ductus arteriosus pointed by white arrow (B), bicuspid aortic valve (C) and coarctation of the aorta pointed by white arrow (D).



Figure 6: Magnetic resonance images showing the bicuspid aortic valve with lateralized sigmoids - white asterisks, (A), coarctation of the aorta (white arrow) and persistence of ductus arteriosus pointed by white asterisk (B).

as left of the anterior leaflet, unicuspid mitral valve, prolapse of the anterior mitral leaflet and mitral supralvalvular membrane [10,11]. Other studies have also found abnormalities of the mitral valve. Our cases have been asymptomatic for a long time, it is possible that the mitral valve abnormalities may progress with time and result in worse condition of the patients. The second case developed symptoms in early stage because the involvement of the atrioventricular valves, the aortic valve and aorta.

Conclusions

On the basis of two cases we can conclude that when Ebstein's anomaly is diagnosed it is mandatory to investigate what happens in the left side of the heart, because in the literature there are reports about the presence of different lesions of the left heart associated with Ebstein's anomaly of the tricuspid valve. The non-invasive imaging techniques play a very important role in the diagnosis, prognosis and treatment of these patients. An understanding of the morphological

and functional details of Ebstein's anomaly is the most important point for the correct interpretation of the noninvasive images and adequate clinical judgment. Finally, it is mandatory to assess the left cavities of the heart in all patients with Ebstein's anomaly, in order to establish a complete evaluation.

Consent

„Written informed consent was obtained from the patient for publication of this case report and any accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal.“

Author Contributions

NEZ performed and interpreted the echocardiographic studies, participated in the design of the study, in the anatomic-echocardiographic correlation and helped to draft the manuscript and made the translation from Spanish into English. GHP participated in the design of the study and in the writing and translation of the manuscript from Spanish into English. GMR performed and interpreted the magnetic resonance images. LMC made the dissections and photographs of the heart with Ebstein's anomaly and participated in the anatomic-echocardiographic correlation.

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