Ebstein's Anomaly of the Tricuspid Valve

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A 27-year-old female presented with three months of palpitations and chest pain. Upon cardiac auscultation, a 3/6 systolic murmur could be heard at the left lower sternal border followed by a loud S3. The systolic murmur increased during inspiration; and no diastolic murmur was appreciated. The electrocardiogram revealed sinus rhythm, right bundle branch block and right-axis-deviation (**Figure 1A**). A 15-day cardiac monitoring demonstrated only rare premature ventricular contractions. A chest radiograph performed during the initial visit demonstrated an enlarged right heart silhouette, a rectified left heart border due to a displaced and dilated right ventricle and a small ascending aortic knob (**Figure 1B**), A transthoracic echo (**Figure 1C**) revealed severely dilated right heart chambers, decreased right ventricular systolic function and an apically displaced septal leaflet of the tricuspid valve with

Figure 1A. Electrocardiogram revealing right bundle branch block and heart axis deviation to the right.

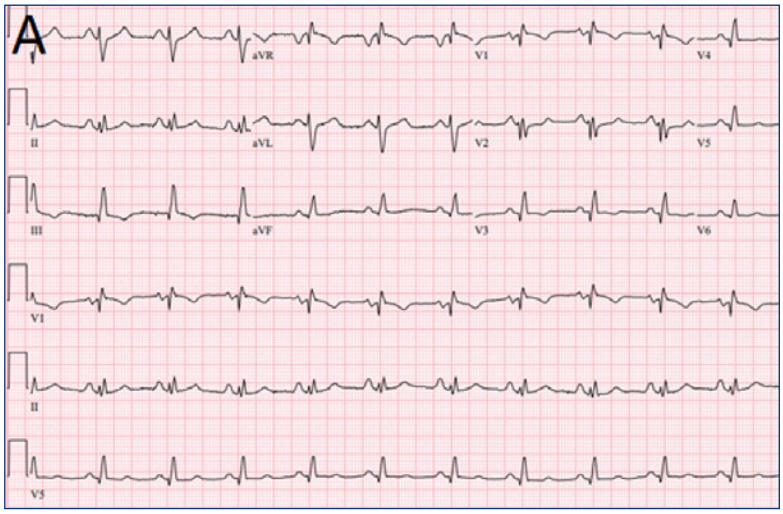


Figure 1B: Chest radiograph

Chest radiograph demonstrates an enlarged right atrium making up the entire right heart border (1), straightening of the left heart border (2) and a small ascending aortic knob (3). The cardiac silhouette is also enlarged with a cardiothoracic ratio greater than 50% on a PA view of the chest (4).

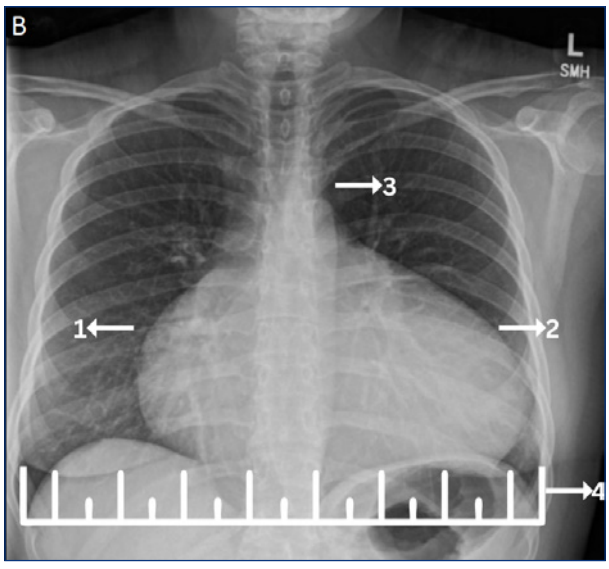


Figure 1C: Transthoracic Echocardiogram

Transthoracic echocardiogram showed a markedly dilated right atrium with an "atrialized" right ventricle and apical displacement of the septal leaflet of the tricuspid valve, characteristic of Ebstein's anomaly. In addition, there was a small left ventricle (LV) and left atrium (LA) with a flattened interventricular septum, suggesting pressure and volume overload. No other congenital abnormalities were identified.

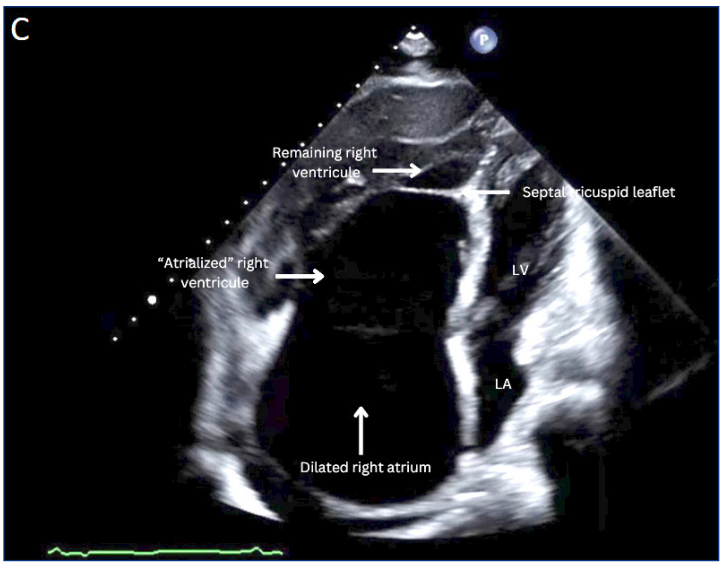
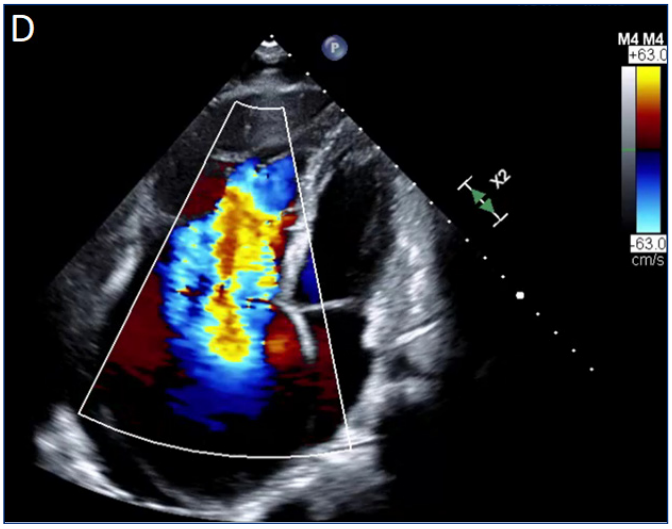


Figure 1D. Transthoracic Echocardiogram – doppler of tricuspid valve
Doppler of the tricuspid valve revealed severe tricuspid regurgitation.



"atrialization" of the right ventricle. There was severe tricuspid valve regurgitation (**Figure 1D**), also seen on cardiac MRI, which confirmed regurgitant fraction of 78%, but no evidence of intracardiac shunting. Finally, for functional capacity evaluation, the patient underwent cardiopulmonary exercise testing which was non-diagnostic for ischemia due to failure to achieve 85% of age-predicted maximum heart rate likely in the setting of betablocker therapy started before the study. The test demonstrated impaired exercise capacity based on peak VO2 of 19.2 ml/kg/min (47% predicted). Surprisingly, our patient lived almost three decades without any major symptoms that would have precipitated prior investigation.

Despite the significant anatomic disruption seen in the images, she presented clinically with no overt signs or symptoms of heart failure. After shared decision making, patient was managed conservatively with betablockers for symptom control. She was referred to cardiothoracic surgery for close surveillance of any changes in clinical status that would warrant anatomic correction.

This constellation of findings is consistent with Ebstein's anomaly; a congenital malformation of the tricuspid valve with a wide spectrum of presentations depending on the degree of anatomic disruption. Patients with minimally displaced leaflets, or less impaired ventricular function, may develop appropriately until adulthood and remain asymptomatic or only mildly symptomatic. In contrast, the disease may also manifest with severe cardiomegaly (often seen in a routine anteroposterior chest radiograph), right heart failure, pulmonary hypoplasia or even intracardiac shunts. The impact of such severe pathophysiologic changes translates into the high rates of perinatal morbidity and mortality in the affected population. In addition, conduction diseases such as accessory pathways and Wolf-Parkinson-White are strongly associated with Ebstein's anomaly, potentially leading to malignant arrhythmias and sudden cardiac death.

Finally, regarding treatment strategies, the approach may vary from symptom relief therapy, aiming for right ventricle unload, to anatomic correction. Surgery is warranted for high degrees of tricuspid regurgitation and right heart failure, severe pulmonary hypertension, intra-cardiac shunts leading to persistent hypoxemia and cyanosis or uncontrolled arrythmias. A negative impact in functionality may also qualify patients for surgical repair, especially considering that most of these individuals are children or young adults at risk for impaired development or limited lifestyle. Pacemakers or implantable cardiac defibrillators might also be indicated to manage or prevent life threatening arrhythmias.

In summary, Ebstein's anomaly is a congenital disease of the tricuspid valve and right heart that can present with a myriad of signs and symptoms. Diagnosis is usually made in the early stages of life prompting surgical correction, or, alternatively, requiring aggressive clinical treatment to ensure a healthy development and lifestyle. Multidisciplinary team and specialized services are also fundamental to provide optimal care and surveillance, mitigating further complications and high rates of mortality.

References

1. Attenhofer Jost CH, Connolly HM, Dearani JA, Edwards WD, Danielson GK. Ebstein's anomaly. Circulation. 2007 Jan 16;115(2):277–85.
2. Celermajer DS, Bull C, Till JA, et al. Ebstein's anomaly: presentation and outcome from fetus to adult. J Am Coll Cardiol. 1994;23:170.
3. Zuberbuhler JR, Allwork SP, Anderson RH. The spectrum of Ebstein's anomaly of the tricuspid valve. J Thorac Cardiovasc Surg. 1979;77:202–211.

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Disclosures

The submitting author has received a consent form from the participant involved in this study. The study fulfilled all local requirements for the "research ethics board" and principles of the COPE have been reviewed. The authors have no conflicts of interest to disclose.

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