

Case Report

When Preoperative Echocardiography Reveals More Than Expected: an Incidental Diagnosis of Ebstein Anomaly

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Abstract

Background: Ebstein anomaly is an uncommon congenital cardiac defect characterized by the abnormal apical displacement of the septal and posterior leaflets of the tricuspid valve, leading to atrialization of a portion of the right ventricle. The clinical spectrum is highly variable, ranging from severe cyanosis and heart failure in the neonatal period to mild or asymptomatic disease that may remain undiagnosed until adulthood. Although some patients remain clinically stable for years, incidental detection of Ebstein anomaly in adults is relatively uncommon.

Case Presentation: A middle-aged patient presented with a traumatic greater trochanteric fracture and was scheduled for orthopedic surgery. As part of routine preoperative evaluation, transthoracic echocardiography was requested. The study unexpectedly revealed marked apical displacement of the septal tricuspid leaflet, severe right atrial enlargement, atrialized right ventricle, and moderate-to-severe tricuspid regurgitation consistent with Ebstein anomaly. The patient denied previous cardiac symptoms, cyanosis, syncope, or documented arrhythmias. Electrocardiography demonstrated sinus rhythm without pre-excitation. Following multidisciplinary discussion, orthopedic management was modified and the patient was referred for congenital cardiology evaluation.

Conclusion: This case highlights the importance of comprehensive cardiovascular assessment before non-cardiac surgery and illustrates how congenital heart disease may remain undiagnosed until adulthood. Incidental recognition of Ebstein anomaly can significantly alter perioperative management and long-term follow-up.

Keywords: Ebstein Anomaly, Congenital Heart Disease, Preoperative Echocardiography, Tricuspid Regurgitation, Incidental Diagnosis.

INTRODUCTION

Ebstein anomaly is a rare congenital cardiac malformation, accounting for less than 1% of all congenital heart defects and occurring in approximately 1 in 20,000 live births (1). The underlying embryological abnormality results from incomplete delamination of the tricuspid valve leaflets from the right ventricular myocardium during fetal development. Consequently, the septal and posterior (inferior) leaflets remain abnormally attached to the right ventricular wall and become displaced apically toward the right ventricular apex, producing the characteristic anatomical features of Ebstein anomaly (2). It has highly variable clinical presentation. Severe forms may be diagnosed prenatally or during the neonatal and childhood periods, (1) particularly in the presence of severe tricuspid regurgitation,

cyanosis, heart failure, functional pulmonary atresia, or associated congenital cardiac abnormalities (3). In contrast, milder forms may remain asymptomatic for many years and consequently escape detection until adulthood. The 2025 American Heart Association/American College of Cardiology guidelines highlight that some individuals with mild Ebstein anomaly may remain asymptomatic and be diagnosed incidentally as late as the sixth or seventh decade of life (4). This broad clinical spectrum emphasizes that Ebstein anomaly may remain unrecognized until adulthood and may occasionally be identified incidentally during evaluation for an unrelated medical or surgical condition. The clinical course and prognosis of Ebstein anomaly are highly variable and depend on the degree of tricuspid valve malformation, severity of tricuspid regurgitation, right

ventricular dysfunction, associated intracardiac shunts, and the presence of arrhythmias. Major complications include progressive right-sided heart failure, severe tricuspid regurgitation, right atrial and right ventricular dilatation, cyanosis due to right-to-left shunting across an atrial septal defect or patent foramen ovale, paradoxical embolism, infective endocarditis, and both atrial and ventricular arrhythmias. Accessory atrioventricular pathways and Wolff–Parkinson–White syndrome are particularly associated with Ebstein anomaly and may result in recurrent supraventricular tachycardia or, rarely, sudden cardiac death (5). The impact on quality-of-life ranges from minimal limitation in asymptomatic individuals to substantial exercise intolerance, fatigue, heart failure, recurrent arrhythmias, and reduced functional capacity in patients with advanced disease. Management is individualized according to symptoms, tricuspid regurgitation, right ventricular size and function, cyanosis, and arrhythmic burden (6). Hence, Ebstein anomaly is a congenital disease that has a broad range of clinical manifestations and even wider range of treatment modalities from wait and see to GDMT and Surgical intervention.

Case Presentation

A 65-year-old male presented to the emergency department after a fall resulting in a greater trochanteric fracture. There was no prior history of congenital heart disease, cyanotic episodes, palpitations, syncope, or heart failure symptoms. Physical examination demonstrated stable hemodynamics. Cardiovascular examination revealed a soft pansystolic murmur at the left lower sternal border. Oxygen saturation was normal on room air. Routine preoperative investigations showed nothing significant except for the fact his electrocardiogram showed Right Bundle Branch Block. For which transthoracic echocardiography was performed. Echocardiography demonstrated Apical displacement of the septal tricuspid leaflet exceeding 8 mm/m² body surface area, markedly enlarged right atrium, Atrialization of the proximal right ventricle, Redundant anterior tricuspid leaflet, Moderate-to-severe tricuspid regurgitation, Preserved left ventricular systolic function, No significant pulmonary hypertension, No pericardial effusion. These findings established the diagnosis of Ebstein anomaly that was later confirmed by Cardiac MRI.

The patient remained asymptomatic from a cardiovascular perspective. Orthopedic and cardiology teams jointly assessed perioperative risk. Surgical planning was modified with attention to arrhythmia surveillance, volume management, and anesthetic considerations. The patient subsequently underwent fracture management without major cardiovascular complications.

DISCUSSION

Ebstein anomaly is an uncommon congenital cardiac malformation characterized by abnormal formation and apical displacement of the tricuspid valve, resulting in atrialization of a variable portion of the right ventricle and functional reduction of the right ventricular chamber. Although traditionally considered primarily a tricuspid valve abnormality, contemporary understanding recognizes Ebstein anomaly as a more extensive right ventricular cardiomyopathy that may also affect left ventricular structure and function (1). The 2025 American College of Cardiology/American Heart Association (ACC/AHA) guideline (4) estimates its occurrence at approximately 1 in 1,000 live births in contemporary series. The anatomical and haemodynamic spectrum is broad, ranging from severe disease presenting in fetal life or infancy to minimally symptomatic disease that remains undiagnosed until adulthood. The European Society of Cardiology (ESC) (6) similarly emphasizes that the clinical consequences depend not only on the degree of tricuspid valve displacement, but also on the severity of tricuspid regurgitation, extent of right ventricular atrialization, functional right ventricular size and contractility, associated intracardiac shunts, and arrhythmias.

The present case is clinically significant because Ebstein anomaly was detected incidentally during preoperative cardiovascular evaluation for a greater trochanteric fracture in a patient with no previously known history of congenital heart disease. This case highlights the considerable phenotypic variability of Ebstein anomaly and demonstrates that milder forms of the condition may remain asymptomatic and undiagnosed until adulthood. Importantly, the absence of overt cardiac symptoms in adults does not exclude clinically significant disease, as progressive tricuspid regurgitation, right atrial and right ventricular enlargement, deterioration of right ventricular systolic function, right-to-left shunting at the atrial level, impaired exercise tolerance, and atrial or accessory-pathway-mediated tachyarrhythmias

may develop with advancing disease. According to ESC guidelines, atrial septal defect or patent foramen ovale and accessory pathways are among the most commonly associated abnormalities in Ebstein anomaly (6). Furthermore, the coexistence of multiple accessory pathways and atrial tachyarrhythmias may increase the risk of sudden cardiac death.

The echocardiographic diagnosis (figure 1) in this patient is particularly important because the hallmark anatomical feature of Ebstein anomaly is apical displacement of the septal and posterior tricuspid valve leaflets, with an enlarged and often sail-like anterior leaflet (7). This displacement produces an atrialized segment of the right ventricle proximal to the functional tricuspid valve. The resulting malcoaptation of the abnormal tricuspid valve frequently produces tricuspid regurgitation, the severity of which is a major determinant of clinical outcome. In adult patients, assessment should therefore extend beyond confirmation of leaflet displacement to include quantification of tricuspid regurgitation, right atrial and right ventricular dimensions, functional right ventricular volume and systolic function, presence and direction of an atrial-level shunt, pulmonary pressures, oxygen saturation, and assessment for associated congenital lesions. The 2025 ACC/AHA guideline (4) recommends multimodality assessment of tricuspid morphology, right-heart size and right ventricular function, together with rhythm assessment, pulse oximetry and functional assessment.

Cardiac magnetic resonance imaging has an important complementary role when echocardiography does not adequately characterize the anatomy or quantify right ventricular involvement (8). The 2025 ACC/AHA guideline states that CMR can be useful in adults with Ebstein anomaly for assessment of anatomy, right ventricular dimensions and systolic function, particularly when these measurements are required for management or surgical planning. This is particularly relevant because accurate assessment of the functional and atrialized portions of the right ventricle may be difficult using conventional two-dimensional echocardiography. In stable patients, the guideline also supports periodic CMR surveillance according to anatomical and physiological severity, with shorter intervals in patients with moderate or severe tricuspid regurgitation or right ventricular dysfunction.

An important clinical implication of an incidental diagnosis is the potential for complications despite the absence of symptoms. Arrhythmias are among the most important long-term complications of Ebstein anomaly (9). Accessory atrioventricular pathways are relatively common, and approximately one-third of adults with ventricular pre-excitation may have multiple accessory pathways, which are associated with a higher risk of sudden cardiac death. Atrial tachyarrhythmias are also common in adults with Ebstein anomaly. The 2025 ACC/AHA guideline (4) therefore recommends electrophysiological assessment in patients with symptomatic arrhythmia or asymptomatic ventricular pre-excitation to identify high-risk or multiple accessory pathways. Catheter ablation is recommended for high-risk accessory pathway conduction or multiple accessory pathways, and electrophysiological assessment before tricuspid valve surgery may be useful because surgical intervention can make subsequent catheter access to right-sided arrhythmogenic substrates more difficult.

The diagnosis also has particular relevance in the setting of non-cardiac surgery. The 2025 ACC/AHA guideline (4) emphasizes that adults with moderate or complex congenital heart disease undergoing cardiac or non-cardiac procedures should have involvement of an adult congenital heart disease specialist to assist with procedural, anaesthetic and post-procedural management. In a patient with Ebstein anomaly, perioperative assessment should consider right ventricular reserve, severity of tricuspid regurgitation, volume status, oxygenation, pulmonary pressures and the presence of arrhythmogenic substrates. Avoidance of significant changes in preload and afterload and careful management of fluid balance are particularly important in patients with substantial right-sided disease (10). Thus, the incidental echocardiographic diagnosis in the present case was clinically relevant not merely as an anatomical finding but because it potentially altered perioperative risk assessment and multidisciplinary planning.

Management of Ebstein anomaly in adults is individualized according to symptoms, tricuspid regurgitation, right ventricular size and function, cyanosis, exercise capacity and arrhythmic burden. Patients with mild disease, preserved ventricular function and no significant symptoms or complications may be managed conservatively with lifelong follow-up through an adult congenital heart disease

service. The 2025 ACC/AHA guideline recommends regular clinical assessment, ECG and transthoracic echocardiography, with the frequency determined by the patient's physiological stage. More advanced disease requires closer surveillance and additional imaging or functional testing. The ESC guideline similarly recommends management in experienced congenital heart disease centres because of the anatomical complexity and variable natural history of this lesion.

Surgical intervention is primarily directed toward significant tricuspid valve dysfunction and its consequences. According to the 2025 ACC/AHA guideline, surgery is recommended in adults with at least moderate tricuspid regurgitation when accompanied by heart failure, worsening exercise capacity or progressive right ventricular systolic dysfunction.

The present case therefore highlights an important aspect of adult congenital cardiology clinically silent congenital disease may be discovered incidentally during evaluation for an unrelated condition, yet the diagnosis has implications extending beyond the immediate clinical encounter. Identification of Ebstein anomaly permits assessment of the patient's long-term risk of progressive tricuspid

regurgitation, right ventricular dysfunction, cyanosis, paradoxical embolism and arrhythmias, while also allowing appropriate perioperative risk stratification. In an asymptomatic adult without significant tricuspid regurgitation, right ventricular dysfunction, cyanosis or arrhythmia, conservative surveillance may be appropriate; however, the patient should remain under lifelong adult congenital heart disease follow-up. The present case reinforces the value of careful preoperative echocardiographic assessment and demonstrates how an incidental finding can lead to recognition of a previously undiagnosed congenital cardiac condition with important implications for both perioperative care and long-term cardiovascular management.

CONCLUSION

Ebstein anomaly may remain clinically silent until adulthood and can be discovered incidentally during routine preoperative workup. Echocardiography plays a crucial role in identifying this rare congenital condition. Recognition before non-cardiac surgery facilitates risk stratification, perioperative optimization, and long-term cardiology follow-up.

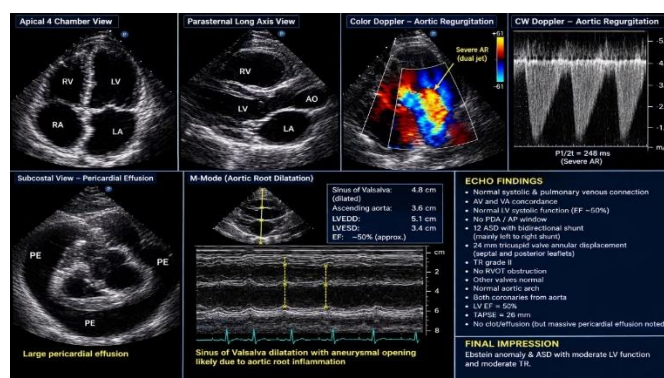


Figure 1: Detail Echocardiographic Study of the Patient

REFERENCES

1. Pasqualin G, Boccellino A, Chessa M, Cicone G, Marcolin C, Micaglio E, et al. Ebstein's anomaly in children and adults: multidisciplinary insights into imaging and therapy. *Heart*. 2024;110(4):235-244. doi:10.1136/heartjnl-2023-322420.
2. Ebstein anomaly. In: StatPearls [Internet]. Treasure Island (FL): StatPearls Publishing; [cited 2026 Sep 8]. Available from: [NCBI Bookshelf](#)
3. Cueto-Robledo G, Yañez-Marquez MF, Garcia-Lezama M, Ramos-Pacheco VH, Gonzalez-Hermosillo LM, Mendoza-Lopez AC, et al. Ebstein anomaly: imaging, pathophysiology, and therapeutics. *Cureus*. 2026;18(7):e112612. doi:10.7759/cureus.112612.
4. 2025 ACC/AHA/HRS/ISACHD/SCAI Guideline for the Management of Adults With Congenital Heart Disease]. *J Am Coll Cardiol*. 2025. doi:10.1016/j.jacc.2025.09.006. [JACC guideline article](#)
5. Eckerström F, Hjortdal VE, Eriksson P, Dellborg M, Mandalenakis Z. Morbidity burden in patients with Ebstein anomaly: the natural history. *J Am Heart Assoc*.

2024;13(21):e037127.

doi:10.1161/JAHA.124.037127.

6. Habib G, et al. 2020 ESC Guidelines for the management of adult congenital heart disease. Eur Heart J. 2021;42(6):563-645. doi:10.1093/eurheartj/ehaa554. [ESC Adult Congenital Heart Disease Guidelines](#)
7. Dalakoti M, Singh D, Yeo WT, Tay LWE, Poh KK. Electrocardiography findings and clinical presentation in Ebstein's anomaly. Singapore Med J. 2019;60(11):560-565. doi:10.11622/smedj.2019146.
8. Yalonetsky S, Tobler D, Greutmann M, Crean AM, Wintersperger BJ, Nguyen ET, et al. Cardiac magnetic resonance imaging and the assessment of Ebstein anomaly in adults. Am J Cardiol. 2011;107(5):767-773. doi:10.1016/j.amjcard.2010.10.058.
9. Gupta A, Prabhu MA, Anderson RD, Prasad SBV, Campbell T, Turnbull S, et al. Ebstein's anomaly: an electrophysiological perspective. J Interv Card Electrophysiol. 2024;67(4):887-900. doi:10.1007/s10840-024-01744-8.
10. Praxis Medica. Article 55 [Internet]. [cited 2026 Sep 8]. Available from: Praxis Medica – Article 55