VOLUME - 14, ISSUE - 07, JULY - 2025 • PRINT ISSN No. 2277 - 8160 • DOI : 10.36106/gjra

Original Research Paper

Medicine

CASE REPORT: EBSTEIN ANOMALY IN A MIDDLE-AGED WOMAN

Dr. Ankur Rana MD, Microbiology, Medical Officer Specialist, Government of Himachal Pradesh, India.

ABSTRACT A 45-year-old woman presented with progressive dyspnea and palpitations, revealing a rare case of Ebstein anomaly diagnosed in adulthood. Echocardiography showed apical displacement of the tricuspid valve, severe tricuspid regurgitation, and a dilated right atrium. Managed with furosemide and metoprolol, she was referred for surgical evaluation (cone procedure). This case highlights the importance of considering congenital heart defects in adults with unexplained right heart dysfunction and underscores the role of echocardiography in reducing diagnostic delays.

KEYWORDS : Ebstein anomaly, congenital heart disease, tricuspid regurgitation, right heart failure, adult diagnosis.

INTRODUCTION

Ebstein anomaly, a rare congenital heart defect (prevalence: 1 in 200,000 live births), is characterized by apical displacement of the tricuspid valve leaflets, leading to right atrial enlargement, tricuspid regurgitation, and arrhythmias. While often diagnosed in childhood, milder forms may remain undetected until adulthood, posing diagnostic challenges due to atypical presentations. This report describes a delayed diagnosis of Ebstein anomaly in a middle-aged woman, emphasising clinical features, diagnostic tools, and management strategies.

Case Presentation

Patient Information:

Mrs. Anita, a 45-year-old woman employed in a chemical packaging unit in the industrial area of Kala Amb, Himachal Pradesh, presented with a 6-month history of worsening exertional dyspnea (NYHA Class II–III) and intermittent palpitations. She reported that a heart murmur was noted during childhood, but no cardiac investigations were pursued due to limited healthcare access and financial constraints. She is a non-smoker and does not consume alcohol. Her obstetric history includes three full-term vaginal deliveries, all uneventful. There is no known family history of congenital heart disease.

Clinical Examination:

- Vital Signs: Blood pressure 118/76 mmHg, heart rate 82 bpm, oxygen saturation 98% on room air.
- Cardiovascular Findings: Jugular venous distension, a holosystolic murmur at the left lower sternal border, and a widely split first heart sound (S1). No cyanosis or peripheral edema.
- Respiratory System: Clear lung fields, no crepitations or wheeze.

Investigations:

- Electrocardiogram (ECG): Right bundle branch block, right atrial enlargement (P-wave amplitude >2.5 mm in lead II), and intermittent premature atrial contractions.
- Echocardiogram: Apical displacement of the septal tricuspid leaflet by 12 mm/m^2 (Celermajer index: grade 2), severe tricuspid regurgitation, markedly dilated right atrium. No atrial septal defect was seen.
- Cardiac MRI: Confirmed atrialization of the right ventricle with reduced right ventricular ejection fraction (RVEF) of 35%. No associated structural defects were noted.
- NT-proBNP: Mildly elevated at 428 pg/mL.
- 6-minute walk test: Reduced endurance, total distance covered: 330 meters.

Management And Referral:

Mrs. Anita was started on oral furosemide (20 mg daily) and metoprolol (25 mg twice daily) for symptom control and arrhythmia prevention. Given her progressive symptoms and structural severity, she was referred to a tertiary cardiac center in Chandigarh for further evaluation and planned cone procedure for tricuspid valve repair. She was advised to limit heavy physical labor at work and follow up regularly for reassessment.

DISCUSSION

Ebstein anomaly in adults is increasingly recognized due to advances in cardiac imaging. Diagnostic clues include right heart enlargement and characteristic echocardiographic findings, such as apical displacement of the tricuspid valve and an atrialized right ventricle. Mrs. Anita's delayed diagnosis likely stemmed from a mild phenotype, with childhood murmurs overlooked due to limited access to advanced imaging. Differential diagnoses, including pulmonary hypertension and right ventricular cardiomyopathy, were excluded based on normal pulmonary artery pressures on echocardiography and characteristic MRI findings.

Adults with Ebstein anomaly often present with arrhythmias (20–30% have Wolff-Parkinson-White syndrome) or heart failure symptoms. Mrs. Anita's palpitations and NYHA Class II–III symptoms align with this pattern. Medical management focuses on symptom relief with diuretics and beta-blockers, while surgical options, such as the cone procedure, offer favorable outcomes, with 5-year survival rates exceeding 80% in adults [1]. The 2015 American Heart Association/American College of Cardiology (AHA/ACC) guidelines recommend lifelong follow-up at specialized congenital heart disease centers [2]. This case underscores the need for heightened awareness of congenital anomalies in adults with unexplained right heart symptoms.

CONCLUSION

This case highlights Ebstein anomaly as a critical differential in adults with right heart dysfunction. Early echocardiography is essential for timely diagnosis. Multidisciplinary care, including surgical planning at specialized centers, optimizes outcomes. Increased awareness of atypical presentations and routine cardiac screening in patients with unexplained symptoms could reduce diagnostic delays and improve prognosis.

Consent

Written informed consent was obtained from the patient for publication.

REFERENCES

1. Attenhofer Jost CH, Connolly HM, Dearani JA, et al. Ebstein's anomaly. Circulation. 2007;115(2):277-285.
2. Stout KK, Daniels CJ, Aboulhosn JA, et al. 2015 AHA/ACC guideline for the management of adults with congenital heart disease. J Am Coll Cardiol. 2015;67(23):e157-e185.
3. Dearani JA, Mora BN, Nelson TJ, et al. Ebstein anomaly: surgical management and outcomes. Ann Thorac Surg. 2019;108(1):122-131.