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Case Report

A case of Ebstein's anomaly in an adult female

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ABSTRACT

Ebstein's anomaly is a congenital cyanotic heart disease of the tricuspid valve and right ventricle, often diagnosed in childhood but can occasionally be present in adulthood with variable clinical manifestations. Here, we report the case of a 25-year-old female with no prior cardiac history who presented with progressive exertional dyspnoea, palpitations, and intermittent dizziness for a year. Subsequently, the baseline and diagnostic investigations were done and the patient was diagnosed with Ebstein's anomaly with severe tricuspid regurgitation. The patient was started on medical management but due to persistent symptoms surgical correction was done. There was a significant improvement in her symptoms thereafter. Hence this case highlights the importance of considering congenital heart diseases as a cause of heart failure in adults.

Keywords: Ebstein's anomaly, Cone reconstruction, Adult congenital heart disease, Tricuspid regurgitation

INTRODUCTION

Ebstein's anomaly is a rare cyanotic congenital heart disease due to a defect in the development of the tricuspid valve resulting in reduced pulmonary blood flow.[1]

According to Harrison's Principles of Internal Medicine, the abnormal development of the tricuspid valve results in: apical and posterior displacement of the dilated tricuspid valve annulus, dilation of the "atrialized" portion of the right ventricle, fenestrations, redundancy, and tethering of the anterior leaflet of the tricuspid valve.[2]

The tricuspid valve deformity can be regurgitant or stenotic in nature.[2]

According to Sharma et al Ebstein's anomaly accounts for 0.3% to 0.6% of all congenital heart defects and is reported to occur in 0.2 to 0.7 per 10,000 live births.[3,4]

Carpentier et al proposed the following classification of Ebstein's anomaly: type A, anterior leaflet is freely mobile, septal and posterior leaflets are moderately displaced and volume of the right ventricle is adequate; type B, anterior leaflet is freely mobile, septal and posterior leaflets are markedly displaced and volume of the right ventricle is reduced; type C, anterior leaflet has restricted mobility due to partial adhesions to the ventricular free wall, septal and posterior leaflets are markedly displaced and hypoplastic and both volume and contractility of the right ventricle is diminished; type D, anterior leaflet has adhesions with the infundibulum, the ventricular free wall and the septal and posterior leaflets, giving a "tricuspid sac" appearance.[5] The right ventricular wall is thin and poorly contractile.

In this case report, we present a case of Ebstein's anomaly in a young female patient.

CASE REPORT

A 25-year-old female presented to us in the outpatient department with complaints of progressive exertional dyspnoea, palpitations, and occasional dizziness over the past year. She had no significant past medical history, and her childhood development was unremarkable without any

episodes of cyanosis, heart murmurs, or exercise intolerance. She worked as a school teacher and led an active lifestyle until her symptoms began affecting her daily activities.

On examination, the patient was alert and oriented. Vital signs revealed a heart rate of 110 bpm, blood pressure of 110/70 mmHg, respiratory rate of 18 breaths per minute, and oxygen saturation of 92% on room air. On general examination there was 2+ pedal edema, but no cyanosis, clubbing, icterus, pallor or lymphadenopathy. The internal jugular veins were distended with prominent "a" wave.

Cardiac auscultation revealed a widely split-second heart sound and a holosystolic murmur best heard at the left lower sternal border. The rest of the systemic examination was unremarkable.

An electrocardiogram (ECG) demonstrated sinus tachycardia, right atrial enlargement, and incomplete right bundle branch block (RBBB). Chest X-ray showed significant cardiomegaly. Echocardiography revealed apical displacement of the septal tricuspid leaflet (>20 mm/m^2 indexed to body surface area), severe tricuspid regurgitation, and atrialization of the right ventricle. The functional right ventricle was significantly reduced in size but exhibited preserved contractility.

The laboratory investigations done are summarised in the following tables:

Table 1: Haematological parameters.

Parameters	Value	Reference range
Haemoglobin (Hb)	14.0 g/dl	13.5-17.5 g/dl (men), 12.0-15.5 g/dl (women)
Haematocrit (Hct)	42%	38-50% (men), 36-46% (women)
Red blood cell count (RBC)	5.0×10^6/µl	$4.5\text{-}5.9\times10^6$/µl (men), $4.1\text{-}5.1\times10^6$/µl (women)
White blood cell count (WBC)	7.0×10^3/µl	$4.0\text{-}11.0\times10^3$/µl
Platelet count	300×10^3/µl	$150\text{-}450\times10^3$/µl
Mean corpuscular volume (MCV)	90 fl	80-100 fl
Mean corpuscular haemoglobin (MCH)	30 pg	27-34 pg

Table 2: Serum chemistries.

Parameters	Value	Reference range
Sodium (Na^+)	140 mmol/L	135-145 mmol/l
Potassium (K^+)	4.0 mmol/L	3.5-5.0 mmol/l
Chloride (Cl^-)	102 mmol/L	98-106 mmol/
Bicarbonate (HCO_3^-)	26 mmol/L	22-29 mmol/l
Blood urea nitrogen (BUN)	15 mg/dl	7-20 mg/dl
Creatinine	1.0 mg/dl	0.7-1.2 mg/dl (men), 0.5-1.1 mg/dl (women)
Glucose	90 mg/dl	70-99 mg/dl (fasting)
Calcium (Ca^{2+})	9.5 mg/dl	8.5-10.2 mg/dl
Magnesium (Mg^{2+})	2.0 mg/dl	1.7-2.2 mg/dl
Phosphate	3.5 mg/dl	2.5-4.5 mg/dl

Table 3: Liver function tests.

Parameters	Value	Reference range
Alanine aminotransferase	30 U/l	7-56 U/l
Aspartate aminotransferase	25 U/l	10-40 U/l
Alkaline phosphatase	80 U/l	44-147 U/l
Total bilirubin	0.8 mg/dl	0.1-1.2 mg/dl
Albumin	4.2 g/dl	3.5-5.0 g/dl

Table 4: Thyroid function tests.

Parameters	Value	Reference range
Thyroid-stimulating hormone	2.5 mIU/l	0.4-4.0 mIU/l
Free thyroxine (Free T4)	1.2 ng/dl	0.8-1.8 ng/d
Free triiodothyronine (Free T3)	3.0 pg/l	2.3-4.2 pg/ml

Table 5: Cardiac biomarkers.

Parameters	Value	Reference range
NT-proBNP	1000 pg/ml	<125 pg/ml (age <75), <450 pg/ml (age ≥75)
Troponin	0.01 ng/ml	<0.04 ng/ml (assay-dependent)

Table 6: Others.

Parameters	Value	Reference range
C-reactive protein (CRP)	5 mg/l	<10 mg/l
Erythrocyte sedimentation rate (ESR)	10 mm/hr	0-20 mm/hr (men), 0-30 mm/hr (women)

NT-pro BNP levels were elevated, consistent with volume overload.

Based on clinical findings and imaging studies, a diagnosis of Ebstein's anomaly was confirmed. Patient's condition was classified as Carpentier type C (markedly atrialized right ventricle with severely restricted leaflet mobility).

Initial management focused on symptomatic relief and optimizing hemodynamic:

Diuretics

Furosemide was administered to alleviate peripheral oedema and reduce right atrial pressure.

Beta-blockers

Carvedilol was prescribed to control palpitations and improve exercise tolerance.

Counselling

The patient was educated about her condition and advised to avoid strenuous activities that could exacerbate symptoms.

Given her persistent symptoms and significant tricuspid regurgitation, the patient was referred for surgical evaluation. After multidisciplinary discussions, she underwent a cone reconstruction procedure, a surgical technique designed to restore tricuspid valve competence and optimize right ventricular function. The surgery was successful, with intraoperative findings confirming severe tricuspid valve deformity and extensive atrialization of the right ventricle.

The patient's recovery was uneventful. Post-operative echocardiography demonstrated minimal residual tricuspid regurgitation, improved right ventricular function, and resolution of right atrial enlargement. She was discharged on postoperative day 7 with instructions for gradual resumption of physical activity and regular follow-up visits.

At the 6-month follow-up, the patient reported significant improvement in her quality of life and functional capacity, achieving a New York heart association (NYHA) class I status. Repeat echocardiography confirmed durable surgical results with no progression of regurgitation or ventricular dysfunction.

DISCUSSION

Ebstein's anomaly has an unknown aetiology, mostly sporadic in nature. Familial cases are a result of the duplication of chromosome 15q which results in abnormal development of the tricuspid valve. Most cases are asymptomatic. If the deformity is severe, patients will present with heart failure, cyanosis being the most common presentation in neonates.[6]

This case is notable as the patient was diagnosed in early adulthood as opposed to early childhood in most cases.

The most common symptoms are palpitations, dyspnoea, and signs of heart failure like bilateral lower limb swelling. Some patients may have a loud first heart sound, the 'sail sound' due to closure of the anterior leaflet.[7]

In our case, the patient presented with progressive exertional dyspnoea, palpitations, and pedal oedema. On auscultation, split second heart sound was appreciated.

Ebstein's anomaly is typically diagnosed with an electrocardiogram, which reveals RBBB in most cases including ours. Cases may also show tall p waves, atrial fibrillation, flutter, or ectopic atrial tachycardia.[8,9] Due to the right atrial enlargement, patients might develop cardiomegaly, which can be visualised on chest x-ray.[10]

In this case, the patient's chest x-ray showed significant cardiomegaly with a globular-shaped heart and decreased pulmonary vascular markings.

For prenatal detection, a 2-dimensional echocardiogram (2D-ECHO) is the diagnostic tool of choice. It is also used in adults to visualise the size of the heart chambers.[9] First line management is conservative, aiming at correcting heart failure. Tachyarrhythmias are corrected by radiofrequency ablation of the accessory pathways. Surgery is indicated in symptomatic adults unresponsive to medical treatment. Surgical procedures include tricuspid valve repair or replacement, single ventricle

repair and in cases with severe left ventricular dysfunction, heart transplantation.[10]

In our case, the patient was initially started on medical treatment and counselled about surgical treatment modalities. She underwent a cone reconstruction surgery which successfully improved her condition.

CONCLUSION

Ebstein's Anomaly has an idiopathic aetiology. Our patient was diagnosed based on clinical findings and imaging studies. This case highlights the presence of Ebstein's anomaly of type C Carpentier classification (markedly atrialized right ventricle with severely restricted leaflet mobility).

In this case, prompt diagnosis and initial medical treatment followed by surgical treatment led to significant improvement in the patient's quality of life and functional capacity.

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