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Case report of Ebstein's anomaly in a young female

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Abstract

Ebstein's anomaly, also known as Ebstein's malformation, is a congenital heart defect that occurs in about 0.005% of live births and accounts for 0.3–0.6% of all congenital heart disease. It is due to delamination failure at embryologic state, or displacement of the leaflets from the ventricular myocardium. In people with Ebstein's abnormality, the tricuspid valve does not close properly, leading to regurgitation. Here is the case report of a 24-year-old female patient who was diagnosed with Ebstein's anomaly 5 years back and had been on follow-up. However, she missed appointments and discontinued the medication for 3 months. She presented with shortness of breath for 5 days, which occurred at rest, and generalized body weakness. Since she was in critical condition, she was admitted to the intensive care unit of the hospital. Subsequently, baseline and diagnostic investigations were done. The health care team initiated immediate treatment, and all available treatments were administered, and the patient's condition improved. In this report, the first presentation of the woman was during adulthood and it was with heart failure even though most patients with Ebstein's anomaly present during the early age of their life and with arrhythmia. Therefore, the main aim of this case report is to show the atypical presentation of Ebstein's anomaly.

Keywords

Ebstein's anomaly, congenital heart disease, tricuspid valve regurgitation

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Introduction

Ebstein's anomaly is a rare congenital cardiac disease.[1] It is characterized by significant apical displacement of the tricuspid valve, resulting in regurgitation, dilatation of the right atrium and right ventricle, and atrial and ventricular arrhythmias.[2] Hence, the leaflets of the tricuspid valve are deformed and become displaced due to this disorder.[3]

It is described by the abnormal insertion of one or two flaps of the tricuspid valve. While the anterior leaflet is placed in the right position, the septal and the posterior leaflets are affected by this misplacement. This causes displacement of the functional orifice of the tricuspid toward the right ventricular apex or the pulmonary infundibulum, with arterialization of the right ventricle and a decrement of its volume.[3]

Rarely does tricuspid stenosis occur, and in almost all cases tricuspid regurgitation is detected. As a result, Ebstein's abnormality can cause cardiac hypertrophy and heart failure.[4]

The true prevalence of this heart defect is unknown because mild versions are commonly untreated. An echocardiogram is used to diagnose Ebstein's anomaly. It also helps identify any additional heart defects. This test allows the pediatric cardiologist to determine the degree of valve displacement, the severity of valve leakage (insufficiency), or valve narrowing (stenosis). Widespread utilization of echocardiography is intensifying the detection of more cases nowadays.[5]

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Researchers are not sure exactly what increases the risk of a baby having an Ebstein's anomaly. Genetics and environmental factors are believed to be involved. Using some medicines during pregnancy, such as lithium, might increase the risk of Ebstein's anomaly in the child. Maternal exposure to second-hand cigarette smoke at home and a family history of congenital heart disease was associated with elevated odds of Ebstein's anomaly. Genetic analyses could clarify the potential heritability of Ebstein's anomaly.[6] Early age at presentation was frequently associated with other cardiac lesions, particularly atrial septal defect, and pulmonary stenosis, which predisposes to cyanosis from right-to-left shunting.

The clinical presentation varied with age at diagnosis. Fetuses: an abnormal routine prenatal scan (86%), Neonates: cyanosis (74%), Infants: heart failure (43%), Children: Murmur (63%), Adolescents and adults: Arrhythmia (42%).[7] But in the case of our patient who is in the adult age group, the presentation is heart failure rather than arrhythmia. Unlike the common presentation of Ebstein's anomaly, this patient presented during adulthood and with heart failure.

Case summary

This is a 24-year-old female known as Ebstein's anomaly patient for the past 5 years. She was on furosemide 20 mg, which she has been taking two times a day after she presented with symptoms and signs of heart failure. She had follow-ups, but she was not adherent and missed appointments. Besides, she discontinued her medications for the last 3 months due to financial reasons. Currently, she is presented with a chief complaint of shortness of breath at rest of 5 days duration. Associated with this, she has a worsening cough, which is productive with whitish sputum estimated as half an Arabic coffee cup per day, low-grade intermittent fever, poor appetite, and fatigue of 5 days duration. She also has orthopnea of two pillows, paroxysmal nocturnal dyspnea of 3 weeks duration, and palpitations of 2 weeks duration. She has no family history of known cardiac diseases. She has a history of previous admission to the intensive care unit (ICU) 2 years back with the diagnosis New York Heart Association (NYHA) class 4 stage c congestive heart failure plus unstable ventricular tachycardia (V-tach) with cardiogenic shock plus severe community-acquired pneumonia plus Vitiligo. She presented with shortness of breath at rest, easy fatigability, worsening of palpitation, and restlessness of 2 days duration. She had symptomatic improvement of the heart failure between the admissions, but she had a history of intermittent exertional dyspnea.

On physical examination her vital sign was BP = 90/55, RR = 34, PR = 148 (IR), T° = 36.1°C, SPO2 = 46% at room air.

On the respiratory system, there was a severe sub-costal and inter-costal retraction, coarse crackles over the left posterior basal, and fine crackles over the right basal chest with relatively decreased air entry.

Cardiovascular system examination revealed peripheral pulses are not palpable, but raised jugular venous pressure, hyper-dynamic precordium apical & parasternal heave, and point of maximal impulse is not shifted. Also, grade III HSM (holo systolic murmur) is best heard at the left lower sternal border (LLSB).

Other system findings were unremarkable.

Blood workups: White blood cell counts = 10,500/uL (lymphocytes = 29.1% and neutrophil = 60.4%), hemoglobin = 21.2 g/dL, HCT = 50.9%, MCV = 80.4 fL and platelet was 312×10^3 serum electrolytes (K = 4.61, Na = 137.4, Cl = 101.9, iCa = 1.21, nCa = 1.07, Tca = 2.07, and serum creatinine = 0.9).

On the echocardiography, there is downward displacement of the tricuspid valve with severe tricuspid regurgitation, mild concentric left ventricular hypertrophy, and mild left ventricular systolic dysfunction (ejection fraction = 49%, calculated numerically). It also showed dilated right atrial and right ventricular chambers, mild mitral valve regurgitation, and slightly dilated inferior vena cava with prominent hepatic veins. The patient's left ventricular hypertrophy and left ventricular dysfunction can be explained by the possible shunt that led the patient to develop cyanosis. This shunt could not be specified due to the unavailability of saline echocardiography in our setup, which is one of our limitations and reason for referral (Figure 1).

The EKG also showed irregularly irregular rhythm, no distinct *p* waves, right axis deviation, prominent *R* wave (M shape) on lead *V*1 and W shape on lead *V*6 (right bundle branch block), delta wave on lead III, aVL, *V*1–*V*3.

The patient has pre-excitation since she has a delta wave with wide QRS with tachycardia, but it is difficult to comment on the PR interval since there is no distinct *p* wave.

On chest X-ray, a huge cardiomegaly around the ratio of 0.8–0.9 was found (Figure 2).

Then, the patient was diagnosed with NYHA class IV stage c heart failure secondary to Ebstein's anomaly precipitated by drug discontinuation and severe community-acquired pneumonia. Associated with this, she was also diagnosed with atrial fibrillation with fast ventricular filling. She was on face mask oxygen with re-breather 15 L/min, challenged with Lasix then maintained with 40 mg IV twice a day (after adequate U/O), digoxin 0.25 mg PO four times a day for the first 24 then 0.125 mg PO per day, ceftriaxone 1 gm IV twice a day, and Azithromycin 500 mg PO per day for 3 days, UFH (unfractionated heparin) 5000 IU SBQ twice a day and salt-free diet.

The differential diagnosis for Ebstein's anomaly is Eisenmenger syndrome, congenital tricuspid valve dysplasia with regurgitation, arrhythmogenic right ventricular cardiomyopathy, and atrial septal defect.

The patient indicated surgery since she developed heart failure and cyanosis, the setup, Wachemo University NEMMCS hospital, has diagnostic and treatment limitations to do sub-specialist evaluation and surgical management. As

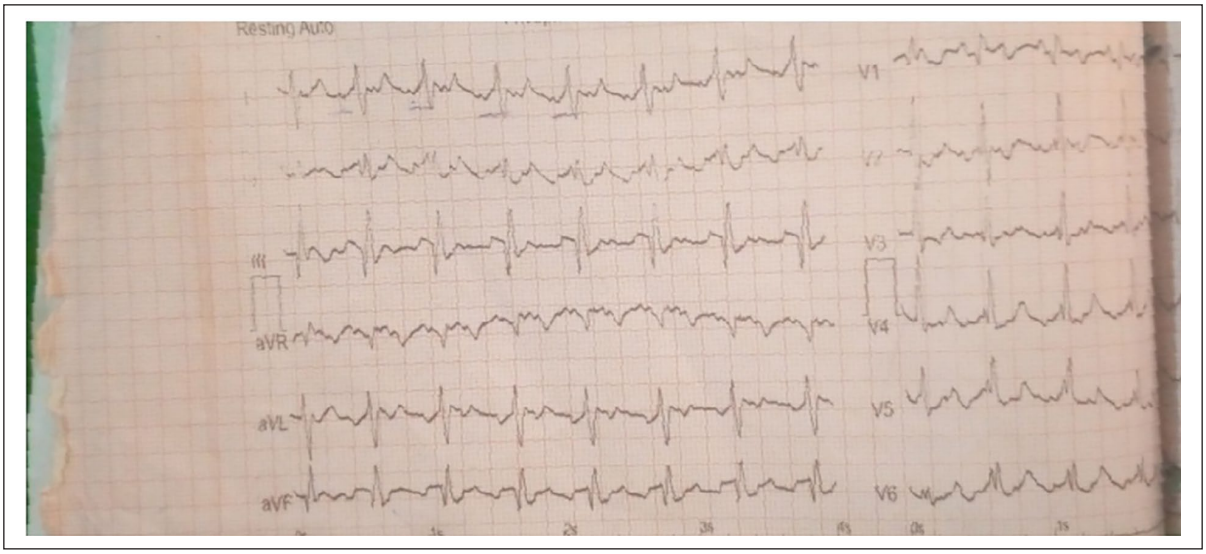

Figure 1. Electrocardiography: indicated sinus rhythm, RBB with WPW (pre-excitation and symptomatic tachyarrhythmia) pattern.
RBB: right bundle branch block; WPW: Wolff Parkinson White Syndrome.

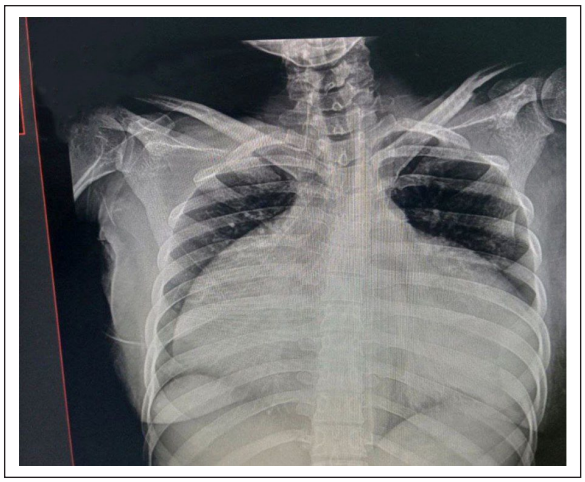

Figure 2. Shows chest X-ray finding of huge cardiomegaly around the ratio of 0.8–0.9.

a result, she was referred to another institution with a better setup for possible cardiologist and cardiothoracic surgeon evaluation and subsequent treatment. Hence, the patient requires surgical intervention since it is the definitive management of the case.

Discussion

Ebstein's anomalies can range from minimal to severe. If the deformity of the tricuspid valve is severe, it may result in profound congestive heart failure in the neonatal period or even death. At the other end of the spectrum, patients with a mild degree of tricuspid displacement and dysfunction, as was the case in our patient, may remain asymptomatic until young adulthood or in some cases late adulthood.[8]

A large spectrum of concomitant lesions has been described. Our patient had mild concentric left ventricular hypertrophy and mild left ventricular systolic dysfunction (ejection fraction = 49%). It also showed dilated right atrial and right ventricular chambers. There is also mild mitral valve regurgitation.

It should be emphasized the therapeutic approach in these patients may be in question sometimes because the operative indication of asymptomatic Ebstein's anomaly in adult patients has not been defined.[2] Operative repair of Ebstein's anomaly is performed, usually during younger age, although medical management may be used to manage some of the symptoms of heart failure and arrhythmias. Observation alone may be advised for asymptomatic and acyanotic patients, with mild cardiomegaly, and normal exercise tolerance. Most patients in NYHA classes I and II can be managed medically. Eventually, most patients will require surgery. Surgery is offered when the patient's symptoms progress to NYHA class III or IV, increasing cyanosis becomes evident, or if paradoxical embolism occurs.[3]

Many surgeons believe that severe tricuspid regurgitation with moderate right ventricular dysfunction can be the operative indication in adult patients with asymptomatic Ebstein's anomaly, especially when tricuspid valve repair is possible.[2] Surgery is also advised if there is evidence of decreasing exercise performance by exercise testing, progressive increase in the heart size on chest radiography, progressive right ventricular dilation or reduction of systolic function by echocardiography, or appearance of atrial or ventricular arrhythmias. In borderline situations, the echocardiographic determination of tricuspid valve repair decides to proceed earlier with surgery easier.[3]

The cardiomegaly can be such that the heart occupies the entire thorax. In Ebstein's disease, the cardiac silhouette on

the thoracic radiography may vary from normal to cardiomegaly, depending on the severity of the disease. A cardiothoracic index >0.65 cm is a predictor of a bad prognosis of the disease. Nine of our patients had clinical manifestations that may be suggestive of Ebstein's heart disease in its severe form. She had stage IV of NYHA dyspnea, cyanosis, atrial fibrillation, extra-systoles, right heart failure, systolic murmur of tricuspid insufficiency, and cardiomegaly with a cardiothoracic index at 0.86 cm.

The diagnosis of Ebstein's disease is based on echo-Doppler or three-dimensional echocardiography, which shows the bundling of the leaflets, the size of the residual functional right ventricle, as well as the site and the degree of tricuspid regurgitation and the feasibility of valve repair are also assessed by echocardiography. The diagnosis is made by measuring the apical displacement of the septal and posterior tricuspid valve leaflets relative to the anterior mitral valve. A displacement of 8 mm or more per body surface area makes the diagnosis.[9] Contrast echocardiogram using saline is helpful to show patent foramen ovale with right-to-left shunting and bubbles in the left ventricle. There are several cardiopathies associated with tricuspid insufficiency and dilatation of the right cavities that may pose a differential diagnosis with Ebstein's disease.[8]

The most common congenital etiology of tricuspid regurgitation associated with dilatation of the right cavities is in Ebstein's diseases followed by tricuspid valve dysplasia[2] and right ventricular myopathy, in particular right ventricular muscle dysplasia, which can cause tricuspid insufficiency.

On the other hand, rheumatic damage of the tricuspid valve[3,4] and tricuspid valve prolapse[4,5] are the acquired abnormalities that can lead to tricuspid regurgitation and may mimic Ebstein's disease. Less common causes of acquired anomalies and secondary regurgitation include traumatic causes,[3,4,10] endocarditis,[3,5] myocardial infarction, carcinoid disease of the heart, radiation therapy, and connective tissue disease.[4] The good clinical and echocardiographic evaluation makes it possible to differentiate between these various pathologies.

Ebstein's anomaly is a congenital cardiac malformation that can pose serious management, and hemodynamic difficulties as well as electrophysiological ones. The formal surgical indications in Ebstein's disease are clinically significant cyanosis, dyspnea stage III or IV of NYHA, heart failure, a cardiothoracic index >0.65 cm on chest radiography, arrhythmia on ECG, onset of left ventricle function alteration and significant tricuspid insufficiency on transthoracic echography.[1] The case of our patient, she had a severe form of Ebstein's disease indicating immediate surgery in front of the global cardiac decompensating requiring depletion by diuretics contrasting with severe hypotension and rhythm disorders with an indication of vasoactive drugs and antiarrhythmic drugs requiring transfer to the cardiac ICU.

The differential diagnosis for Ebstein's anomaly is Eisenmenger syndrome which is the most severe form of congenital shunt-related pulmonary arterial hypertension (HTN) manifested with a triad of systemic to-pulmonary-shunt (ventricular, atrial, or great artery), pulmonary arterial HTN with shunt reversal (right-to-left), hypoxemia with cyanosis.

The other differential diagnosis is congenital tricuspid valve dysplasia with regurgitation, which is a rare condition with malformed valve leaflets but no excessive displacement or tethering of TV leaflets to the RV myocardium. The other one is arrhythmogenic right ventricular cardiomyopathy which is a clinical entity characterized by ventricular arrhythmias and a specific ventricular pathology. Furthermore, the atrial septal defect is also a possible differential diagnosis which is a defect in the atrial septum and associated left to right shunt causing volume overload on the right heart chambers.

Since the patient's condition needs surgical intervention that cannot be done in our setup, the decision was to refer her to a better institution for further management and follow-up.

The limitation of this report is the information about the patient follow-up like post-intervention evaluation is not stated in the case report because she was referred to another institution.

Conclusion

Ebstein's anomaly is a rare diagnosis but should be considered a differential diagnosis even in elderly patients with severe tricuspid regurgitation and predominant right heart failure in the absence of any other explanation. There may be atypical manifestations of this disease like presentation in adulthood and with heart failure as in the case of this patient. There may be several adult survivors of CHDs who may primarily present with other systemic illnesses.

The patient needs to undergo surgery, which is a tricuspid valve repair known as cone surgery. She may also require shunt closure since the patient is cyanotic.

Author contributions

All authors contributed to drafting the case report, approved the final version, agreed to the journal submitted, and agreed to be accountable for all aspects of the work.

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Ethics approval

Our institution does not require ethical approval for reporting individual cases or case series.

Informed consent

Written informed consent was obtained from the patient(s) for their anonymized information to be published in this article.

Patient consent

Informed written consent was obtained from the patient for all case details and images to be reported in the journal.

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