First presentation of Ebstein's anomaly in an 80-year-old female patient and unusual occurrence of psychiatric illness in her male offsprings

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Accepted on
December 15th, 2012

DOI Name
10.3126/jaim.v2i1.7634

Keywords
Cardiac symptoms, Ebstein's anomaly, psychiatric illness

Citation
Shrestha UK, Sapkota S, Thapa P. First presentation of Ebstein's anomaly in an 80-year-old female patient and unusual occurrence of psychiatric illness in her male offsprings. Journal of Advances in Internal Medicine 2013;02(01):21-3.

ABSTRACT

An 80-year-old female Ebstein's anomaly patient presented with palpitation and dyspnea of New York Heart Association class III. This is probably the eldest surviving Ebstein's anomaly patient presenting for the first time with cardiac symptoms at the age of 80 years. She had four sons and two daughters, among whom the first three sons developed psychiatric disorders after 38 years of age, with one son committing suicide because of major depressive episode. This case is unique because of the late age of initial presentation with cardiac problem and the occurrence of psychiatric illness in the male offsprings of the patient.

INTRODUCTION

Ebstein's anomaly is a rare congenital heart disorder with an incidence of 1 per 200,000 live births, accounting for about 0.3 to 0.7% of all cases of congenital heart disease.[1,2] The anomaly was first mentioned by German physician Wilhelm Ebstein in 1866 in a report titled, "Concerning a very rare case of insufficiency of the tricuspid valve caused by a congenital malformation."[3] Ebstein's anomaly is characterized by tricuspid valve abnormality with apical displacement of the septal and posterior leaflets from the atrioventricular annulus into the right ventricle, leading to the 'atrialization' of a portion of the right ventricle.[4] The clinical presentation depends upon the severity of deformity, whether it is mild, moderate or severe; the age of onset of symptoms can also vary from the patient to patient.[4] Only 5% of patients with Ebstein's anomaly survive beyond the fifth decade and the initial presentation by older patients is rare.[5] There are case reports of Ebstein's anomaly in offsprings following consumption of lithium during early pregnancy by mother.[6,7] However, no cases have been reported where children of patient with Ebstein's anomaly have suffered from psychiatric illness. We reported the unusual case of Ebstein's anomaly with the late age of initial presentation and with the occurrence of psychiatric illness in the male offsprings of the patient.

CASE REPORT

An 80-year-old female was admitted to Kaski Sewa Hospital, Pokhara, Nepal in October, 2012 with palpitation and breathlessness of New York Heart Association (NYHA) function class III for 15 days. She had been well until 15 days prior to the admission and had never complained of shortness of breath or swelling of the ankles before; there was no complain of turning into blue with undue efforts. However, the relatives said that she had not been doing any household works for the last 5 years. She was non-smoker and non-alcoholic, and had never been to the hospital for any significant illness in the past. She denied about having any significant psychiatric illness in the past. She had four sons and two daughters. First son was 52 years old, who developed recurrent depressive disorder at the age of 38 years. Second son was 48 years old, who developed schizoaffective disorder at the age of 38 years. Third son committed suicide at the age of 45 years, which could be due to major depressive episode. Fourth son was 36 old years, not showing any major psychiatric illness so far. Two daughters did not have any psychiatric illness. Her all pregnancy periods were uneventful.

Her body mass index was 15.82 Kg/m^2 (body weight 39 Kg, height 157 cm) and body surface area 1.33 m^2. Her physical examination revealed temperature of 98 degree Fahrenheit, irregularly irregular heart rate of 130 beats/minute, blood pressure of 130/80 mm Hg, respiratory rate of 26 breaths/min and Oxygen saturation of 95%. There were elevated jugular venous distention, positive hepatojugular reflux and slight pedal edema. Heart sounds were irregularly irregular with a varying intensity of S1 component. A grade III/VI pansystolic murmur was best heard at the left lower sternal border, which increased in intensity after a deep inspiration. Respiratory system examination revealed resonance to percussion, good air entry with bilateral basilar rales. The remainder of the physical examination was noncontributory.

Chest radiograph (Figure 1) revealed cardiomegaly with globe-shaped cardiac silhouette and normal vascularity of pulmonary fields. Electrocardiogram (ECG) showed atrial fibrillation with the rate of 130 beats/ minute and incomplete right bundle branch block.

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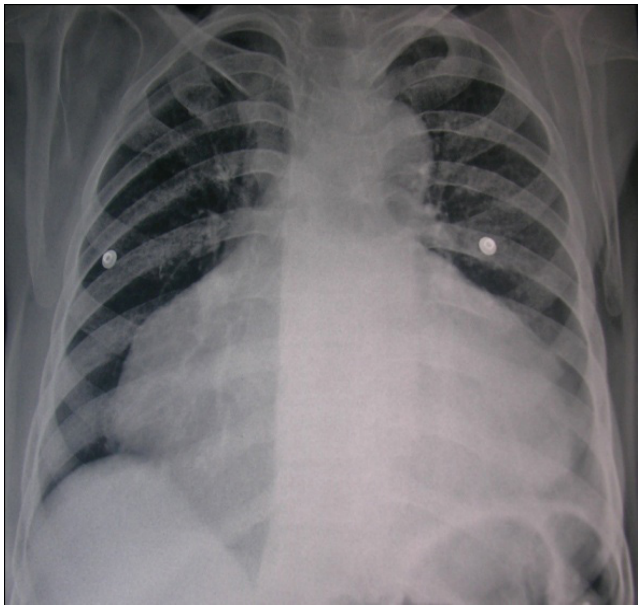

Figure 1: Frontal chest radiograph shows a large heart

Echocardiography (Figure 2) showed apical displacement of septal leaflet of tricuspid valve of about 46 mm from the insertion of anterior leaflet of the mitral valve; this finding was more than 20 mm or 8 mm/m^2 body surface area, the cutoff limit for considering the diagnosis of Ebstein's anomaly.[2,8] Anterior leaflet of tricuspid valve was elongated, redundant, freely mobile and sail-like, with no tethering to the lateral wall of right ventricle. There was no hypoplasia of septal and posterior leaflet. There was marked enlargement of the right atrium and atrialized right ventricle; the combined area of the right atrium and atrialized right ventricle was larger than the combined area of the functional right ventricle, left atrium, and left ventricle measured in the apical four-chamber view at end diastole. There was moderate tricuspid regurgitation with ejection fraction of right and left ventricle 50%. There was no pulmonary artery hypertension and no interatrial communication.The diagnosis of Ebstein's anomaly was made. The anomaly was categorized as of Carpentier type B and Celermajer's extended Glasgow Outcome Scale of Ebstein's anomaly of grade 3.[9,10] Screening Echocardiography of her three surviving sons and two daughters showed normal results.

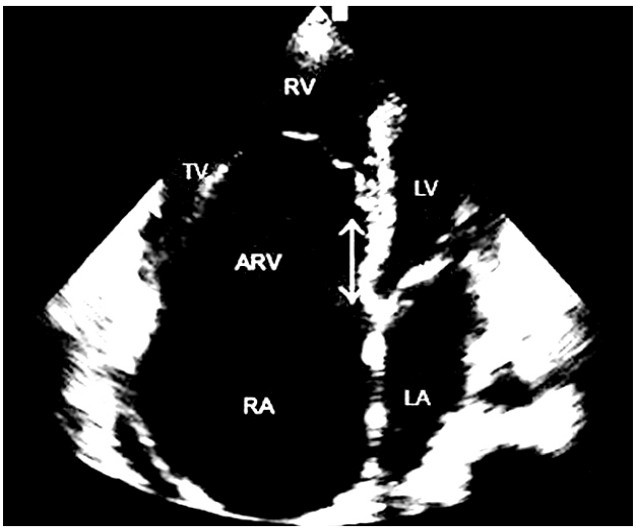

Figure 2: Apical 4-chamber, 2-dimensional echocardiogram shows displacement of the tricuspid valve toward the apex of the right ventricle. TV = tricuspid valve; RV= functional right ventricle; ARV = atrialized right ventricle; RA = right atrium; LA = left atrium; LV = left ventricle.

Laboratory investigations revealed hemoglobin 12.4 gm/dl, total leukocyte count 8,600/cmm (Neutrophil 74% Lymphocyte 24%, Eosinophil 2%), platelet count 220,000/cmm, Erythrocyte Sedimentation Rate 5 mm in first hour, creatine kinase MB 14 U/l, cardiac troponin I negative, blood urea 34 mg/dl, serum creatinine 0.9 mg/dl and random blood sugar 104 mg/dl.

After admission, the patient was treated with diuretics, anticoagulants and digoxin. After 3 days of admission, the fast ventricular rate of atrial fibrillation was controlled. After 4 days of admission, the leg edema and shortness of breath were also improved.

The patient was discharged home after 7 days in a stable condition. She was followed up after 2 months and was doing well with the medications. Her sons were also treated and counseled for their psychiatric illness.

DISCUSSION

The symptoms of Ebstein's anomaly patients are often due to the heart failure caused by the dilated and atrialized right ventricle compressing the left ventricle and, reduced preload for the left heart.[11] The cyanosis, arrhythmia, and sudden cardiac death are the other presenting features.[12] The age at presentation, anatomic severity, hemodynamics, and degree of right-to-left interatrial shunting determine the hemodynamic variations and clinical presentation.

To date, there have been very few reported patients of Ebstein's anomaly surviving longer than 70 years. In 1956, one report was published which described the patient of Ebstein's anomaly, who died at 79 years of age.[13] In 1979, another report was published about Ebstein's anomaly patient, who died at 85 years of age.[14] In 2008, the case was reported about a 77-year-old man with congestive heart failure due to a previously diagnosed condition of Ebstein's anomaly.[4] Recently in 2012, one report was published which described the unusual first presentation of Ebstein's anomaly in a 72-year-old patient.[11]

The patient of our case report was atypical, as she presented to the hospital for the first time at the age of 80 years and had not received any treatment prior to the referral to our hospital. After receiving the treatment with diuretic, digoxin and warfarin, the patient was improved symptomatically. The patient was doing well after a follow up of 2 months without any surgical intervention. Reports suggest the improvement of the patient with Ebstein's anomaly by repair or replacement of the tricuspid valve with plication of the atrialized ventricle.[9,15] However, our patient became symptomatically better and was having NYHA functional class I and atrial fibrillation with controlled ventricular rate even without the surgical intervention.

This is also the first case report describing the occurrence of psychiatric illness in the male offsprings of the Ebstein's anomaly patient. Three consecutive male children of a patient with Ebstein's anomaly were afflicted by a psychiatric illness which started after the age of 38 years. Affective diathesis was predominant, although both schizophrenic and affective components were present in one of the sons. Her only son, who didn't have a psychiatric illness, was younger than 38 years. Both the daughters were spared from the psychiatric illness. The probability that this clustering of late onset of predominantly affective illness affecting only the male offsprings of a patient with Ebstein's anomaly occurred only by chance seems to be low. It, hence, raises the question whether there could be a genetic link between Ebstein's anomaly and affective illness. We are of opinion that this matter is worthy of consideration for further

research. The patient herself could have had episodes of depression of milder variety during her lifetime which was not diagnosed because of lack of mental health services during that time. She might have forgotten those episodes after long duration and we could not find anyone who had known her throughout her life. So, the possibility that this patient might have had both Ebstein's anomaly and an affective disorder has not been conclusively ruled out and this remains the major limitation to correlate Ebstein's anomaly with the psychiatric illness in the male offspring in our case report.

CONCLUSION

In conclusion, the late presentation of Ebstein's anomaly in our patient has shown that the life-span of Ebstein's anomaly patient can be longer and the regular long-term follow up with an experienced cardiologist is required to prevent the further cardiac complications. The regular screening for the psychiatric illness may be needed after 38 years of age in the male offspring of the Ebstein's anomaly patient in order to avoid the uneventful complication of psychiatric illness in the offspring. However, further research is needed to substantiate the association of Ebstein's anomaly patient with the psychiatric illness in the male offspring.

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