



## Case Report of Symptomatic and Asymptomatic Ebstein's Anomaly- Cardiac MR Imaging

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### ABSTRACT

*We hereby report two cases of Ebstein's anomaly, one of the rare congenital heart diseases accounting for only 0.5-1.0 % of congenital heart diseases with incidence of 1 in 2,10,000 live births. Ebstein's anomaly has a wide spectrum of findings, and ages at first presentation while some patients are asymptomatic. We present a classic case of Ebstein's anomaly in a 22-year-old female with massive right sided cardiomegaly, downward displacement of septal tricuspid leaflet, atrialization of right ventricle associated with atrial septal defect. We also present another case of Ebstein's anomaly in a 51-year-old male who was asymptomatic. We also emphasize the role of MRI in assessing apical displacement of the septal leaflet of tricuspid valve, mobility of antero-superior and inferior tricuspid valve leaflets, quantification of tricuspid regurgitation and size of atrialized right ventricle and to assess volume.*

**Keywords-** *Ebstein's anomaly, atrialized right ventricle, tricuspid valve.*

### INTRODUCTION

Ebstein's anomaly of the tricuspid valve, a rare congenital heart defect occurs in approximately 0.5-1% of patients with congenital heart disease. It is characterized by different degrees of dysplasia and the displacement of tricuspid valve leaflets downward into the right ventricle leading to regurgitant anomalous valve.<sup>1</sup> The clinical symptoms in Ebstein's anomaly are mainly right-sided heart failure, arrhythmias, cyanosis and sudden cardiac death. The clinical presentation depends on the age at presentation, the severity of

pathological anatomy, the hemodynamics involved, and lastly the degree of right-to-left inter-atrial shunting.<sup>2</sup> We report two cases of Ebstein's anomaly with the first case presenting in early adulthood with exertional dyspnoea since childhood, and the second case presenting in elderly age who was completely asymptomatic till then. Although the diagnosis of Ebstein's anomaly is usually echocardiographic findings based, its limitations in defining pathologic anatomy in some cases and the advancement of surgical techniques, a complete assessment of the morphologic features is

required which is assessed finely by cardiac magnetic resonance (MR) imaging.<sup>3</sup> We emphasize the role of cardiac MR to accurately assess pathophysiologic changes in Ebstein's anomaly for both morphological and functional aspects.

## CASE REPORT

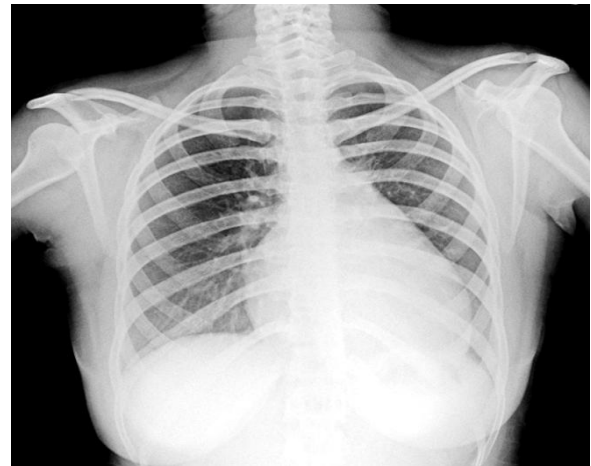
### Case 1 (symptomatic Ebstein's anomaly)

A 22-year-old female had a history of exercise intolerance since childhood. She presented with progressive exertional dyspnoea. Physical examination revealed a holosystolic murmur. The electrocardiography findings included abnormal P waves (indicating right atrial enlargement), a prolonged PR interval and a right bundle-branch block. Echocardiography findings were large "sail-like" tricuspid valve structure within dilated right heart and tricuspid regurgitation. Frontal chest radiograph revealed right sided chamber enlargement with pulmonary oligemia (Fig. 1). Cardiac magnetic resonance (MR) imaging was performed. The findings on both black-blood and bright-blood cine MR images were enlarged right atrium, atrialisation of right ventricle (aRV), downward displacement of tricuspid valve leaflet, leftward bowing of the interventricular septum associated with atrial septal defect (ASD) and minimal pericardial effusion (Fig 2). Cine images viewed as a loop demonstrated regurgitant blood flowing into the right atrium from the right ventricle during systole. The functional RV was reduced in size and function. The stroke volume was 23.9 ml and cardiac output 1.5 l/min. In this case, there was associated ASD which is one of the common association with Ebstein's anomaly.

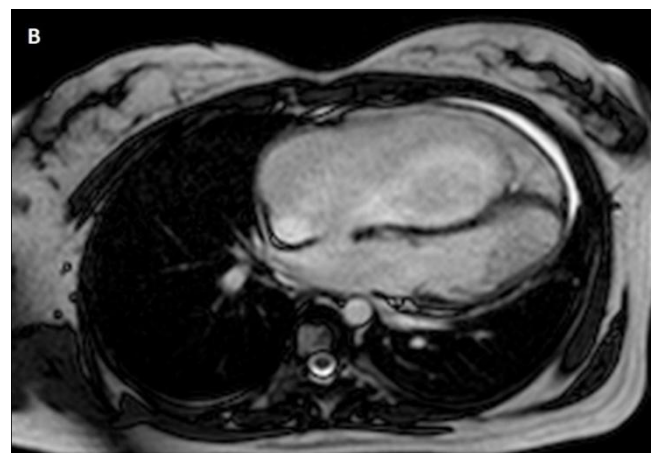
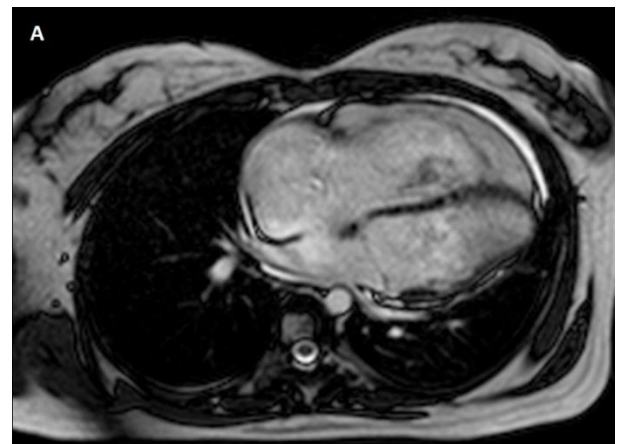
### Case 2 (asymptomatic Ebstein's anomaly):

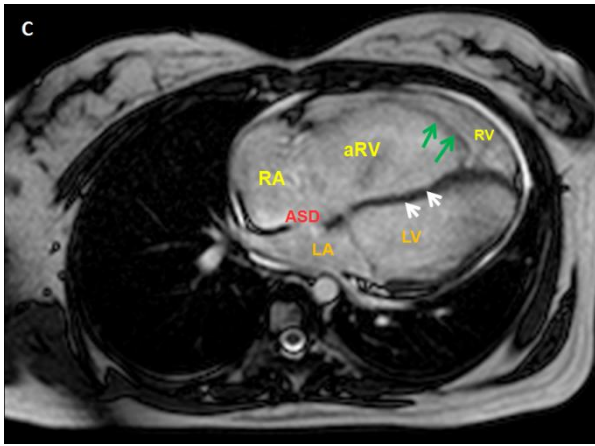
A 51-year-old male who had no symptoms since childhood presented with progressive exertional dyspnea. Electrocardiography demonstrated abnormal P waves, low amplitude QRS waves and prolonged PR interval. Echocardiography findings were large tricuspid valve structure, dilated right heart and tricuspid regurgitation. Cardiac MR imaging showed atrialized right ventricle, downward displacement of tricuspid valve leaflet

(low positioned tricuspid valve), bowing of the interventricular septum, moderate tricuspid regurgitation and minimal pericardial effusion (Figure 3). The left ventricular function was affected by right chamber enlargement and bowing of interventricular septum. The stroke volume was 34.5 ml and cardiac output 2.9 l/min.

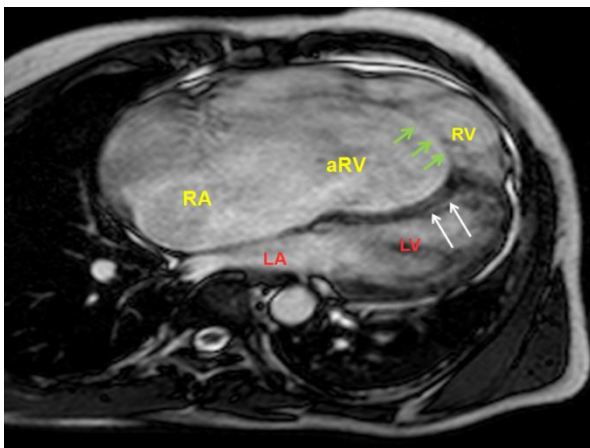


**Fig. 1 :** Postero-anterior chest radiograph of a 22 year-old female with Ebstein's anomaly showing right sided chamber enlargement and pulmonary oligemia.





**Fig. 2 (A-C)** :\_Transverse bright-blood gradient-recalled-echo cine MR images of the heart of a 22 year old female (4-chamber view) with Ebstein's anomaly showing atrialized right ventricle (aRV), downward displacement of tricuspid valve leaflet (Green arrows), leftward bowing of the interventricular septum (White arrow) with atrial septal defect (ASD).



**Fig. 3** :\_Transverse bright-blood gradient-recalled-echo cine MR images of the heart of a 51 year old male (4-chamber view) showing marked downward displacement of shelf-like posterior leaflet of tricuspid valve (Green arrows) with attachment to underlying free wall by numerous muscular stumps, markedly dilated atrialized portion of right ventricle (aRV), small functional portion of right ventricle (RV), leftward bowing of ventricular septum (White arrow) and marked dilatation of right atrium (RA).

## DISCUSSION

These Ebstein's anomaly, a rare congenital heart defect of tricuspid valve occurs in approximately 0.5-1% of patients with congenital heart disease. It is characterized by different degrees of dysplasia

and the displacement of the tricuspid valve leaflets downward into the right ventricle. These anatomic defects divide the right ventricle into a thin walled proximal portion that becomes atrialized and enlarged and a more distal trabeculated component which forms the functional right ventricle.<sup>2</sup>

Ebstein's anomaly is named Wilhelm Ebstein, a German physician. In 1866, Wilhelm Ebstein performed and reported the findings of necropsy in a 19-year-old cyanosed laborer.<sup>4</sup> Although the classical findings are inferior displacement of tricuspid valves and atrialization of right ventricle, the extent of both varies widely. Other factors are leaflet structure, size and extent of adherence to the right ventricle wall, right ventricle contractility and myocardial thickness.<sup>3</sup> Embryogenesis of Ebstein anomaly explains the spectrum of disease. The anterior leaflet develops first embryologically, which arises from the mesenchyme surrounding the atrioventricular orifice and later the posterior and septal leaflets develop through undermining of the myocardium. In Ebstein's anomaly, there is complete or partial failure of undermining of the myocardium, thus leaving the entire septal and posterior valve leaflets and the distal aspect of the anterior valve leaflet low within or adherent to the right ventricle.<sup>5</sup>

Ebstein's anomaly can be a component of complex cardiac diseases or can be associated with ASD, pulmonary stenosis or atresia, mitral stenosis, ventricular septal defect (VSD), corrected or partial transposition of the great vessels and tetralogy of Fallot.<sup>6</sup> In our first case, it was associated with ASD. Because of the variable features of Ebstein anomaly, clinical symptoms vary widely. As a result of congestive heart failure, almost half of the affected untreated patients die during the first year of life.<sup>7</sup> In contrast, in many adult patients the disease is incidentally noted, and are asymptomatic. In our second case, the patient was asymptomatic till presentation. It has varied clinical symptoms including fatigue, dyspnea, and cyanosis, conduction disturbances (Right bundle-branch block and Wolff-Parkinson-White syndrome) and arrhythmias.<sup>8</sup> The presence of cyanosis depends on



the balance between the right and left atrial pressure.

Plain chest radiograph depicts box shaped heart which is caused by right atrial enlargement and pulmonary trunk hypoplasia. Echocardiography shows right heart enlargement, a small functional right ventricle, tricuspid regurgitation and an abnormally downward displaced tricuspid valve. Though the clinical and echocardiographic findings are sufficient to diagnose Ebstein's anomaly, cardiac MR imaging plays a vital role in providing pathological anatomy, morphological features and functional anatomy which in turn helps in planning the surgical technique as it is tailored for each patient.

In our first case, axial bright-blood gradient-recalled-echo cine MR images showed downward displacement of tricuspid valve, atrialisation of right ventricle, reduced functional right ventricle volume, tricuspid regurgitation complicated with atrial septal defect. The size of the functioning right ventricle is reciprocal to the size of the atrialised right ventricle.<sup>3</sup> There is enlargement of the base of the right ventricle caused by displaced tricuspid valve leaflets which ceases to function. The bowing of the inter-ventricular septum is caused by the increase in heart volume in the right side of the heart. In our second case, there was marked downward displacement of posterior leaflet of tricuspid valve with attachment to underlying free wall by numerous muscular stumps, markedly dilated atrialized portion of right ventricle (aRV), marked dilatation of right atrium, small functional portion of right ventricle (RV) and leftward bowing of ventricular septum. No associated congenital heart defects like ASD as seen in the first case. Thus cardiac MR plays a vital role in accurately assessing the pathophysiologic changes.

## CONCLUSION

These two cases of Ebstein's anomaly are presented for their different clinical symptoms which explains the varied clinical presentation of the same. The first case was a young female who was symptomatic while the second case was an elderly male who was asymptomatic till presentation. Though clinical and

echocardiographic findings are sufficient enough to diagnose Ebstein's anomaly, the role of cardiac MR imaging is indispensable in view of providing both morphological and functional anatomy thus helping us in the better understanding of the hemodynamical changes and in the planning of possible surgical techniques.

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