MEDICAL FORUM

# CASE STUDY ON EBSTEIN'S ANOMALY AND THE VALUE OF CARDIAC MRI IN PREOPERATIVE DIAGNOSIS

***Luong Quang Huy*, Nguyen Thi Huyen**, Nguyen Ngoc Trang**, Vu Dang Luu*****

## SUMMARY

Ebstein's anomaly (EA) is a rare congenital heart disease characterized by downward displacement of the septal and inferior tricuspid valve leaflets, redundant anterior leaflets with a sail-like morphology, and atrialization of the inlet portion of the right ventricle. The clinical manifestations of EA are not specific and range from asymptomatic to severe as dictated by the degree of tricuspid valve displacement, the severity of tricuspid regurgitation, effective right ventricular volume, and the presence of associated malformations such as an atrial septal defect, arrhythmias.

We report a case of a 42-year-old female patient with a history of EA diagnosed many years ago, with irregular treatment. Three months prior to admission, the patient began experiencing progressively worsening shortness of breath, leading to hospitalization at Bach Mai Hospital, where she was stabilized with medication and assessed for surgery. The echocardiogram report showed changes in the description of the morphology and attachment sites of the valve leaflets across different examinations. The patient underwent a pre-surgical cardiac magnetic resonance imaging (CMR). She was then surgically treated with the Cone technique for tricuspid valve repair. After surgery, her condition significantly improved.

## I. INTRODUCTION

Ebstein's anomaly (EA) is a rare congenital heart disease characterized by downward displacement of the septal and inferior tricuspid valve leaflets, redundant anterior leaflets with a sail-like morphology, and atrialization of the inlet portion of the right ventricle [1]. The clinical manifestations of EA are not specific, and range from asymptomatic to severe as dictated by the degree of tricuspid valve displacement, the severity of tricuspid regurgitation, effective right ventricular volume, and the presence of associated malformations such as an atrial septal defect, arrhythmias [2].

According to the American College of Cardiology/ American Heart Association (ACC/AHA) Guidelines for the Management of Adults with Congenital Heart Disease [3] determining the anatomy and size of the right atrium and right ventricle in patients with Ebstein anomaly can often be difficult using echocardiography alone, particularly in adults. Cardiac magnetic resonance imaging (CMR) can provide the information necessary for clinical care and surgical planning because CMR data correlates well with intraoperative findings.

We report a case of a 42-year-old female patient with a history of Ebstein›s disease diagnosed many years ago, with irregular treatment. Three months prior to admission, the patient began experiencing progressively worsening shortness of breath, leading to hospitalization at Bach Mai Hospital, where she was stabilized with medication and assessed for surgery. The echocardiogram report showed changes in the description of the morphology and attachment sites of the valve leaflets across different examinations. The patient underwent a pre-surgical

* *Department of Radiology, Hanoi Medical University*

** *Radiology Center, Bach Mai Hospital*

CMR. She was then surgically treated with the Cone technique for tricuspid valve repair. After surgery, her condition significantly improved. Comparison of the post-surgical results with CMR showed a remarkable similarity to the findings observed during surgery.

## II. CASE REPORT

### 1. Clinical presentation

A 42-year-old female patient with a long history of Ebstein's anomaly presented with progressively worsening shortness of breath three months before her admission to Bach Mai Hospital. Upon admission, she exhibited mild shortness of breath, with $SpO_2$ around 95% in room air; regular heartbeat, normal blood pressure, and lung examination were clear with no rales. There were no signs of cyanosis or edema. The patient was subsequently treated medically and stabilized. Following a multidisciplinary consultation, surgery was recommended. She then underwent tricuspid valve repair using the Cone technique, and her condition improved significantly after the operation.

### 2. Paraclinical findings

- Laboratory tests: Basic tests within normal limits.

- ECG: Mildly elevated P waves.

- Chest X-ray (PA view): Cardiomegaly, cardiothoracic ratio 0.61 (Figure 1).

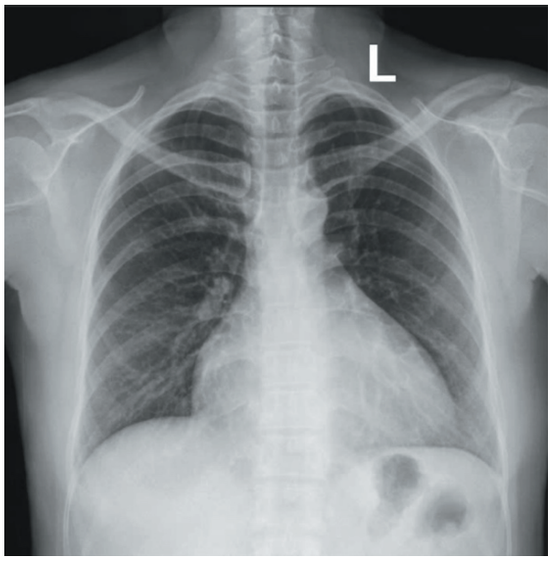



**Figure 1. Chest X-ray showed cardiomegaly with a cardiothoracic ratio of 0.61.**

- Transthoracic echocardiogram (TTE):

+ At the clinic: Ebstein Type C; The septal leaflet of the tricuspid valve was downward displacement, with the distance from the septal leaflet to the anterior leaflet of the mitral valve being 42 mm. The posterior leaflet was atrophied, and adherent to the right ventricular (RV) wall. The anterior leaflet was large, adherent to the left ventricular wall, with limited motion. GOSE (Great Ormond Street Echocardiography) score grade 3. Atrial septal defect with a diameter of 14 mm. Normal left ventricular systolic function.

+ Preoperative TTE: Ebstein Type C; The septal leaflet of the tricuspid valve was downward displacement, with the distance from the septal leaflet to the anterior leaflet of the mitral valve being 22mm. The posterior leaflet had limited mobility and was adherent to the RV wall. The anterior leaflet was small, elongated, mobile, and calcified. GOSE score grade 2. Atrial septal defect with a diameter of 11 mm. Normal left ventricular systolic function.

- CMR was performed by 3T SIGNA Architect (GE Healthcare, USA) (Figure 2) confirmed the diagnosis of Ebstein Type C;

+ The septal leaflet of the tricuspid valve was downward displacement, with the distance from the septal leaflet to the anterior leaflet of the mitral valve being 54 mm. The posterior leaflet was difficult to visualize. The anterior leaflet was elongated, with restricted mobility.

+ Atrial septal defect with a diameter of 8 mm. Normal of the RV outflow tract and pulmonary artery.

+ RV systolic function was approximately 51.44%. Left ventricular systolic function was approximately 58.9%. No late gadolinium enhancement of the myocardium was seen.

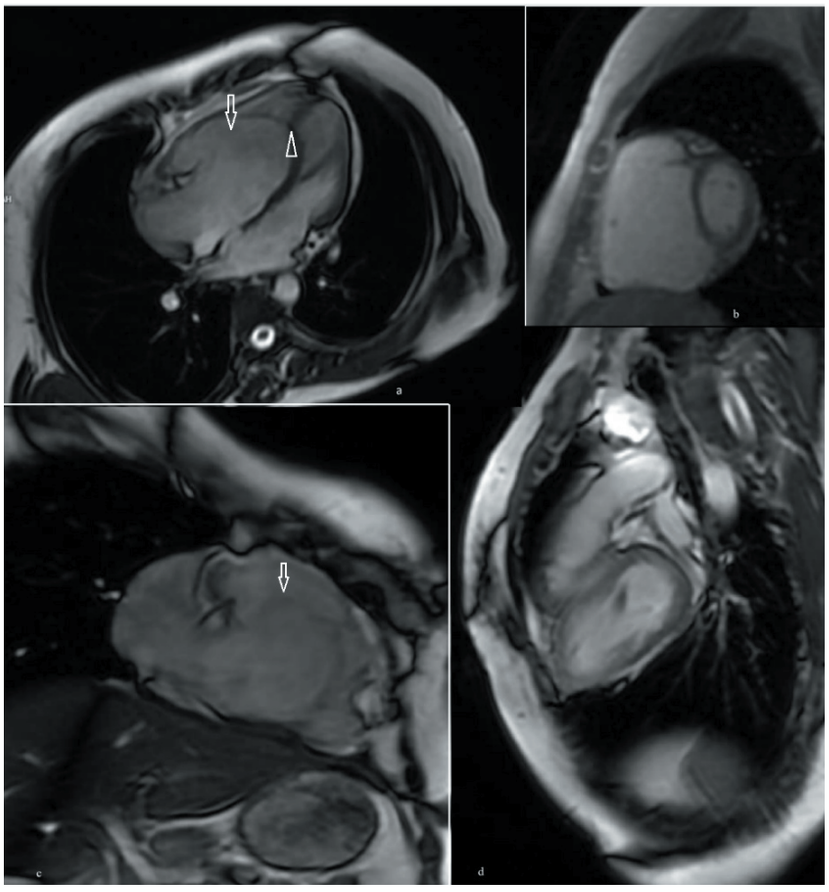
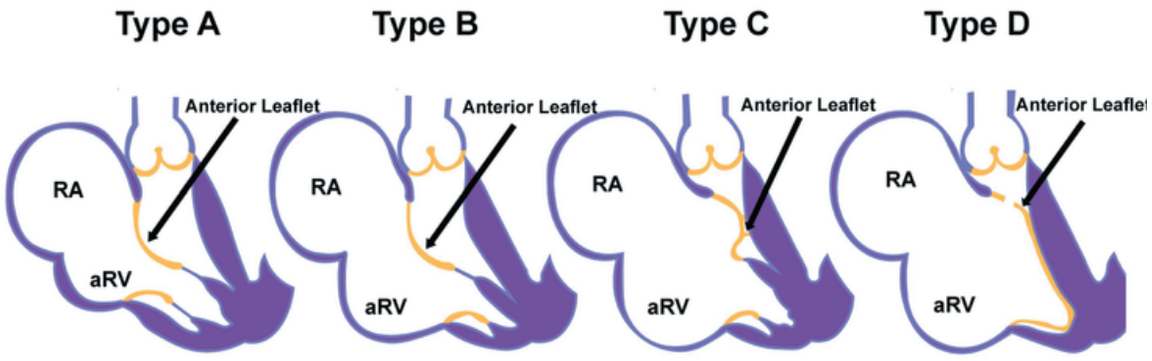


**Figure 2. CMR showed apical displacement of the tricuspid valve (arrowhead in a) with dilatation of the right atrial and atrialized portion of the right ventricle (arrow in a), a redundant anterior leaflet with a sail-like morphology (arrow in c).**

Surgical report: The tricuspid valve exhibited a low attachment, with 45 mm to the true annulus. The septal leaflet was adherent to the interventricular septum. The posterior leaflet was hypoplastic. The anterior leaflet was sail-shaped, with partially restricted motion. The right atrium was moderately dilated, and the atrialized portion of the right ventricle was significantly dilated but remained contractile. Additionally, there was an atrial septal defect measuring 10 mm in diameter.

## III. DISCUSSION

Ebstein anomaly is defined by the following characteristics: apical displacement of the tricuspid valve; adherence of the septal and posterior tricuspid leaflets to the myocardium; apical displacement and dilatation of the tricuspid annulus; dilatation of the atrialized portion of the RV; redundancy, fenestrations, and tethering of the anterior tricuspid leaflet. Regarding classification, in 1988, Carpentier et al. reported the most described morphological classification [4] (figure 3).




| | A | B | C | D |
|---|---|---|---|---|
| Septal/Posterior Leaflet Displacement | Mild | Moderate | Severe | Extremely Severe (Tricuspid Valve Sac) |
| Anterior Leaflet | Normal Morphology and Mobility | Abnormal Attachments with Normal Mobility | Partial Adhesions to the Ventricular Free Wall and Restricted Motion | Severe Adhesions to the Ventricular Wall |
| "Atrialized" Ventricle | Small/Preserved Function | Moderate/Reduced Function | Large/Reduced Function | Very Large/Minimal Function |
| Functional Ventricle | Normal/Preserved Function | Reduced/Preserved Function | Small/Reduced Function | Infundibulum/Depressed Function |

**Figure 3. Carpentier classification of Ebstein's anomaly. RA: right atrium, ARV: atrialized right ventricle; FRV: functional right ventricle [4].**

Abnormalities commonly associated with Ebstein anomaly include a secundum atrial septal defect and a variable degree of RV outflow tract obstruction, etc. The clinical manifestations of EA are not specific and range from asymptomatic to severe. The surgical indication involves considerable debate regarding risk factors and the high mortality rate periprocedural [5]. Therefore, a thorough evaluation is necessary, with complete clinical and paraclinical information.

Traditional imaging modalities for the diagnosis of EA include chest radiography, echocardiography, and cardiac catheterization with right ventriculography. Chest radiography is nonspecific despite occasional pathognomonic appearances. However normal chest radiographic findings do not exclude the diagnosis.

Echocardiography helps confirm the diagnosis, assess the severity of the anatomic lesion, and determine cardiac function [6]. However, it has some disadvantages, including depending on the ultrasound practitioners, can't to provide a panoramic view, and challenges in evaluating right ventricular function [7]. In our case findings, the echocardiogram reports describe the valve lesions and estimate the degree of atrialization, with variations between different ultrasound practitioners. There are limitations in assessing right ventricular function, as well as in evaluating myocardial fibrosis.

CMR has the potential to replace cardiac catheterization for assessing RV function and pulmonary artery anatomy and for providing additional information compared to echocardiography. Some data suggest that CMR and echocardiography should be used as complementary imaging techniques for optimal evaluation before surgery. For example, CMR is preferable when evaluating the right ventricular size and ejection fraction and for visualizing the posterior tricuspid valve leaflet and anterior valve fenestrations, whereas echocardiography may visualize small shunts more readily and may be less likely to underestimate the degree of tricuspid valve regurgitation [8]. CMR can depict anatomy and function with an unrestricted field of view; even the pulmonary arteries and tricuspid valve can be evaluated during a single study [9].

The wide variety of anatomic and pathophysiologic

presentations of Ebstein's anomaly has made it difficult to achieve uniform results with surgical repair, resulting in the development of many different surgical techniques for its repair. In 1993, Da Silva et al. developed a surgical technique involving cone reconstruction of the TV [10]. This operation aims to undo most of the anatomic TV defects that occurred during embryologic development and to create a cone-like structure from all available leaflet tissue. This procedure is illustrated in Figure 4.

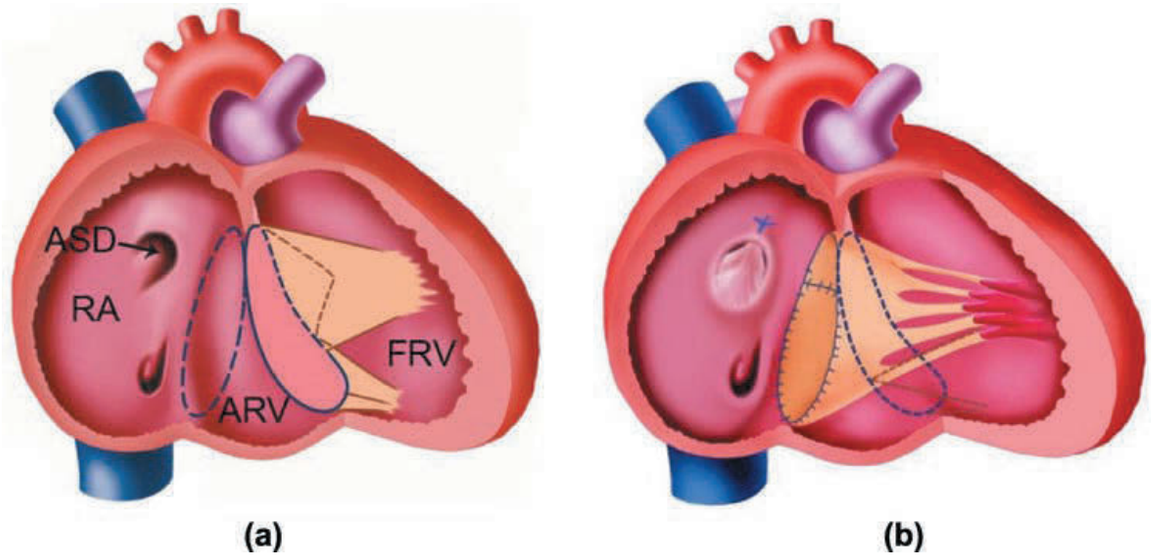



**Figure 4. The cone procedure illustration (b) depicts the tricuspid valve leaflet mobilized and reconstructed in a cone-like shape and reattached to the normal atrioventricular junction, and the atrial septal defect closed in a valved fashion with a single stitch. ASD = atrial septal defect, RA = right atrium, ARV = atrialized right ventricle, functional right ventricle single stitch. ASD = atrial septal defect, RA = right atrium, ARV = atrialized right ventricle, functional right ventricle [10].**

According to a study by Alsaied et al, right ventricle dysfunction is common after a Cone operation for EA and is associated with a higher need for postoperative inotropes. Predictors include lower preoperative right ventricle ejection fraction, a more dilated heart, and more severe tricuspid valve abnormality. The authors noted that preoperative CMR is an important tool in preoperative assessment and helps predict RV dysfunction [11]. Another advantage of CMR is late gadolinium enhancement which is an indicator of myocardial fibrosis, and patients with Ebstein's anomaly frequently demonstrate this finding in the right atrium and right ventricle [12].

In our case, we used a 3T GE MRI, which provides high-resolution imaging of cardiac morphology and function. Aside from the concomitant atrial septal defect, the patient had only Ebstein's anomaly with normal right ventricular function and no late gadolinium enhancement in the myocardium. These are positive prognostic factors that likely contributed to the favorable postoperative outcome and good recovery in our patient.

## IV. CONCLUSION

Ebstein's anomaly is a rare congenital heart disease. The diagnosis is primarily based on echocardiography. However, for preoperative patients, CMR should be indicated to provide detailed information about the heart's morphology and function, as well as a comprehensive view to plan the surgery for the surgeon.

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Correspondent: Luong Quang Huy. Email: quanghuy20591@gmail.com

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