

## Original Article

# Ultrasonic features of fetal ebstein anomaly

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**Abstract:** Objectives: We analyzed and summarized the ultrasonic and anatomical features of fetal Ebstein anomaly to improve the ultrasonographic diagnostic rate. Methods: We analyzed the fetal echocardiographs and either the postnatal echocardiographs or anatomical specimens of 13 cases of fetal Ebstein anomaly. The cases were divided into typical and rare types according to the features of the Ebstein anomaly lesions. Results: Among the 13 cases of Ebstein anomaly examined, 10 cases were typical, and 3 cases were rare types. The typical features of Ebstein anomaly included a downward displacement of the septal and posterior tricuspid leaflets, which were shortened, while the anterior tricuspid leaflet was long. In addition to the typical pathological changes, the rare forms exhibited unique characteristics. The rare forms include an absence of the chordae tendineae and papillary muscle, valve thickening and Ebstein anomaly associated with ventricular noncompaction. Conclusions: Ebstein anomaly is characterized by extensive pathological changes involving the tricuspid valve, chordae tendineae, and papillary and cardiac muscles. According to the features of these lesions, Ebstein anomaly has been divided into typical and rare types. Understanding the ultrasonic features of the different types of Ebstein anomaly can improve the ultrasonographic diagnostic rate of this disease.

**Keywords:** Ebstein anomaly, fetal echocardiography

## Introduction

Ebstein anomaly is a rare and complex congenital heart disease, accounting for less than 1% of all congenital heart diseases and approximately 40% of congenital malformations of the tricuspid valve [1]. There is a high neonatal mortality rate, reaching 85% of affected newborns [2]. Infants with Ebstein anomaly that is diagnosed antenatally have a significantly worse prognosis than those who are diagnosed postnatally. Approximately one-third of infants with Ebstein anomaly die in utero, and only 1/10 survive the neonatal period [3]. Therefore, prenatal diagnosis of the disease has important clinical significance. This study analyzed the fetal echocardiographs and either postnatal echocardiographs or anatomical specimens of 13 cases of fetal Ebstein anomaly. Its features are summarized, categorized. And the diseases are divided into typical and rare types. The aim of our study is to improve the accuracy rate of ultrasound diagnosis.

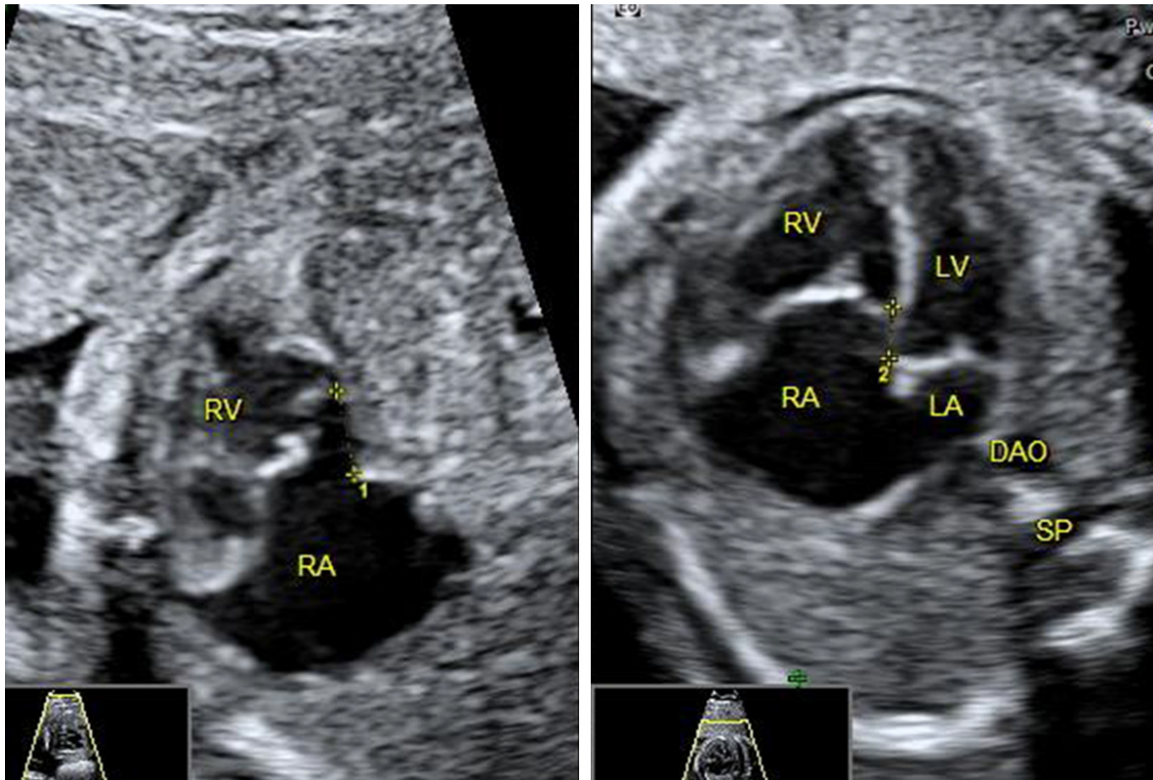
## Materials and methods

Fifty-four fetuses with moderate or severe tricuspid regurgitation at 22-30 weeks of gesta-

tion were recruited from January 2010 to December 2014. The fetuses were examined in the hospital or referred from consultations at other hospitals. Among the 54 cases, 53 were singleton pregnancies, with 1 set of twins. This study was approved through the Institutional Review Board at the medical center, and all the pregnant women provided consent.

Examinations were performed via Voluson E8 (General Electric, USA) with a transabdominal three-dimensional probe (4-8 MHz).

First, we assessed the fetal position and gestational weeks. Then, the upper abdomen transverse, four-chamber, left ventricular outflow tract, right ventricular outflow tract, three-vessel, and three vessels and trachea views were observed using the transverse sweep (sweep technique) [4]. The cardiac axis, cardiac position, cardiothoracic ratio, heart chambers, connection of the atrioventricular valve and ventricular artery, and the echo and movement of each valve were observed. The blood flow through the atrioventricular valve and great vessels was observed using color Doppler flow imaging. The attachment point of the septal and anterior tricuspid leaflets were observed in



**Figure 1.** Ultrasonic image of typical Ebstein anomaly. Abbreviations: LA, left atrium; RA, right atrium; LV, left ventricle; RV, right ventricle; DAO, descending aorta; SP, spine.

the four-chamber view, and the attachment point of the posterior tricuspid leaflet were observed in the two-chamber view.

Ebstein anomaly was diagnosed based on the apical displacement of a tricuspid leaflet and moderate or severe regurgitation, with right atrial enlargement [5]. All cases were followed until birth and confirmed through postnatal echocardiography or autopsy examination after induced labor. Fetuses lost to follow-up were not included in the present study.

In cases of confirmed Ebstein anomaly, postnatal echocardiography and anatomical specimens were analyzed. The ultrasonic features of the chordae tendineae and the papillary and cardiac muscles were evaluated in detail. According to the features of the lesions, the cases were divided into typical and rare types.

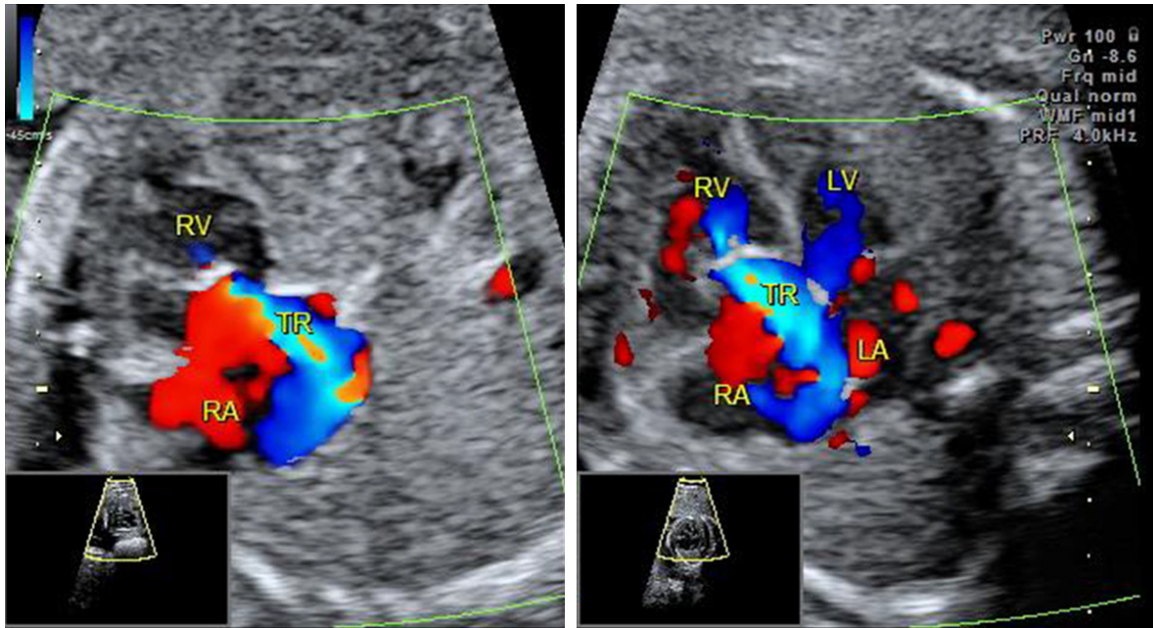
## Results

Among the 54 fetuses with moderate or severe tricuspid regurgitation, 13 fetuses were diagnosed with Ebstein anomaly, and all the cases

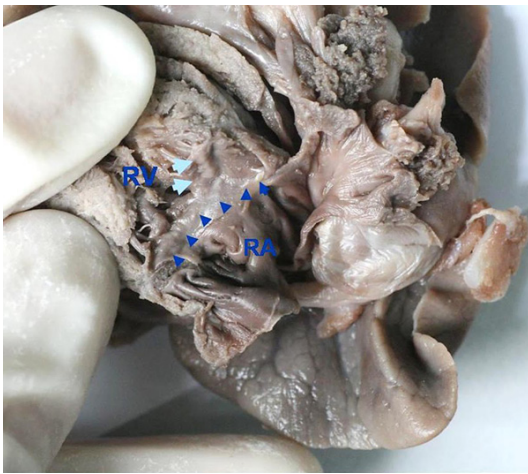
were confirmed through postnatal echocardiography or autopsy. According to the features of chordae tendineae and papillary and cardiac muscles, the cases were divided into typical (10 cases) and rare (3 cases) types. The rare type included an absence of the chordae tendineae and papillary muscle (1 case), valve thickening (1 case) and Ebstein anomaly associated with ventricular noncompaction (1 case). Among the 10 cases of typical Ebstein anomaly, 3 fetuses survived to birth, and 7 pregnancies were terminated. All of the rare-type cases were terminated pregnancies.

### *Ultrasonic and anatomical characteristics of typical Ebstein anomaly*

Three cases of the typical-type Ebstein anomaly survived to birth. The following results of both prenatal and postnatal sonographic examination were obtained: the septal and posterior tricuspid leaflets were shortened and displayed a downward displacement, while the anterior tricuspid leaflet was long and wide with a sail-like motion. The four- and two-chamber views revealed that the right atrium was enlarged,



**Figure 2.** Color Doppler flow imaging of typical Ebstein anomaly. Abbreviations: LA, left atrium; RA, right atrium; LV, left ventricle; RV, right ventricle; TR, tricuspid regurgitation.



**Figure 3.** Anatomical image of typical Ebstein anomaly. Tricuspid valve ring (triangle); apical displacement of the tricuspid valve (arrow). Abbreviations: RA, right atrium; RV, right ventricle.

and an atrialized right ventricle was also observed (**Figure 1**). Color Doppler flow imaging showed moderate or severe tricuspid regurgitation (**Figure 2**). The pregnancies were terminated in 7 of the 10 typical patients. All specimens displayed downward displaced septal and posterior tricuspid leaflets, enlarged right atria, atrialized right ventricles, and shortened chordae tendineae and papillary muscles, supporting the antenatal sonographic diagnosis (**Figure**

**3**). In 9 of 10 typical cases, the fetuses exhibited varying degrees of pulmonary stenosis, and in 1 case, the fetus showed a combined ventricular septal defect without obvious pulmonary stenosis (**Table 1**).

#### *Ultrasonic and anatomical characteristics of rare Ebstein anomaly*

There were 3 cases of rare Ebstein anomaly. One case exhibited chordae tendineae and papillary muscle absence, another case displayed valve thickening, and the last case showed Ebstein anomaly associated with ventricular noncompaction. All of these pregnancies were induced and autopsied. In addition to the typical features of Ebstein anomaly, the antenatal echocardiography and anatomical specimens revealed other unique features (**Table 2**). The case lacking the chordae tendineae and papillary muscle showed that the septal and posterior tricuspid leaflets were directly attached to the ventricular septum and ventricular wall (**Figure 4**). The case with valve thickening exhibited obvious thickening of the tricuspid valve, which was adhered to the ventricular wall (**Figure 5**). The case of Ebstein anomaly associated with ventricular noncompaction showed multiple and prominent myocardial trabeculations and deep intratrabecular recesses woven into a mesh in the ventricular wall. The compact



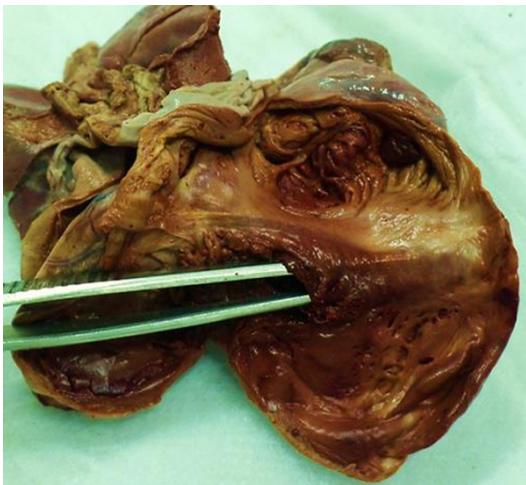
# Prenatal ultrasonography diagnosis of ebstein anomaly

**Table 1.** Ultrasonic and anatomical characteristics of typical Ebstein anomaly

Type	Ultrasonic characteristics (10 cases)	Anatomical characteristics (7 cases)
Typical	Shortened, downwardly displaced septal and posterior tricuspid leaflets	Shortened, downwardly displaced septal and posterior tricuspid leaflets
	Long, wide anterior tricuspid leaflet with a sail-like motion	Shortened chordae tendineae and papillary muscles
	Enlarged right atrium atrialized right ventricle	Long, wide anterior tricuspid leaflet
	Moderate or severe tricuspid regurgitation	Enlarged right atrium atrialized right ventricle
	One case of combined ventricular septal defect and other cases of combined pulmonary stenosis	One case of combined ventricular septal defect and other cases of combined pulmonary stenosis

**Table 2.** Characteristic ultrasound and anatomical features of rare Ebstein anomaly

Type	Characteristic ultrasound and anatomical features
Absence	Lack of chordae tendineae and papillary muscles septal and posterior tricuspid leaflets directly attached to the ventricular septum and the ventricular wall
Thick	Thickened tricuspid valve adherent to the ventricular wall
Noncompaction	Multiple and prominent myocardial trabeculations and deep intratrabecular recesses in the ventricular wall thin, compact myocardium in the outer layer of the ventricular wall, exhibiting an equal or low-level echo



**Figure 4.** Ebstein anomaly associated with absence of the chordae tendineae and papillary muscle. The septal and posterior tricuspid leaflets were directly attached to the ventricular septum and the ventricular wall.

myocardium in the outer layer of the ventricular wall was thinned and exhibited an equal or low-level echo (**Figure 6**).

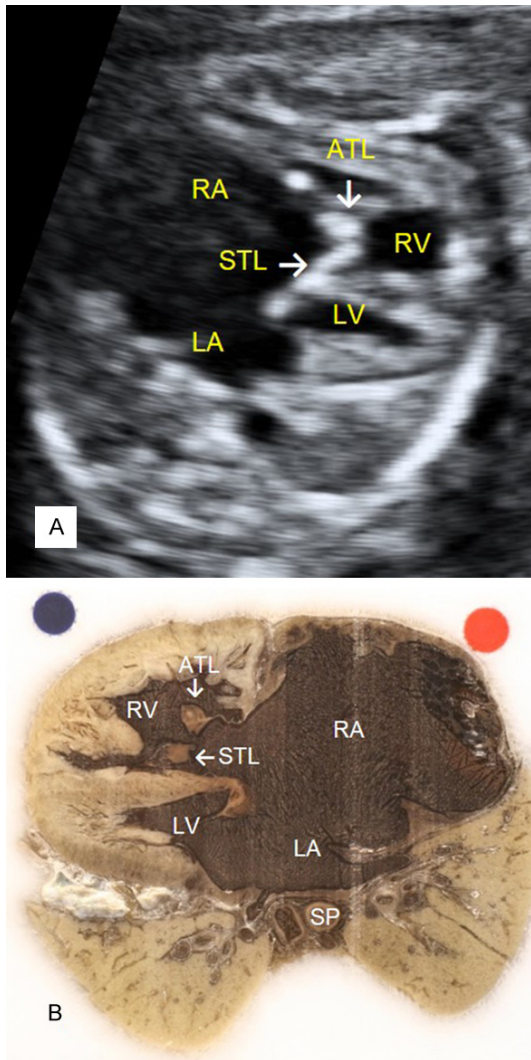
## Discussion

During early embryonic development, the tricuspid leaflet tissue develops from the ventricular wall along with the chordal and papillary muscle apparatus. In Ebstein anomaly, the development of the tricuspid valve from the right ventricle is abnormal, reflecting the obser-

vation of septal and posterior leaflet fusion to the ventricular wall and abnormal, excessive chordal attachments [6]. Thus, Ebstein anomaly is characterized by extensive pathological changes involving the tricuspid valve, chordae tendineae and papillary, and cardiac muscles. The ultrasonic appearances vary according to the different lesions. In contrast with typical Ebstein anomaly, the rare forms include an absence of the chordae tendineae and papillary muscle, valve thickening and Ebstein anomaly associated with ventricular noncompaction. With the development of a precise diagnostic technique, the ultrasonographic diagnostic rate of typical Ebstein anomaly is improving. Thus, it is important to obtain a better understanding of the rare type of this disease to reduce the incidence of misdiagnosis or missed diagnosis.

### *Ebstein anomaly associated with an absence of the chordae tendineae and papillary muscle*

In adult patients, this type of Ebstein anomaly exhibits an absence of the chordae tendineae and papillary muscle and a downward displacement of the septal and posterior tricuspid leaflets [7]. Until recently, this type of Ebstein anomaly had not been reported in fetuses. We observed that the prenatal fetal echocardiography is the same as the typical type. In addition, the anatomical specimen showed downward displacement of the septal and posterior tricuspid leaflets. The septal and posterior tricuspid

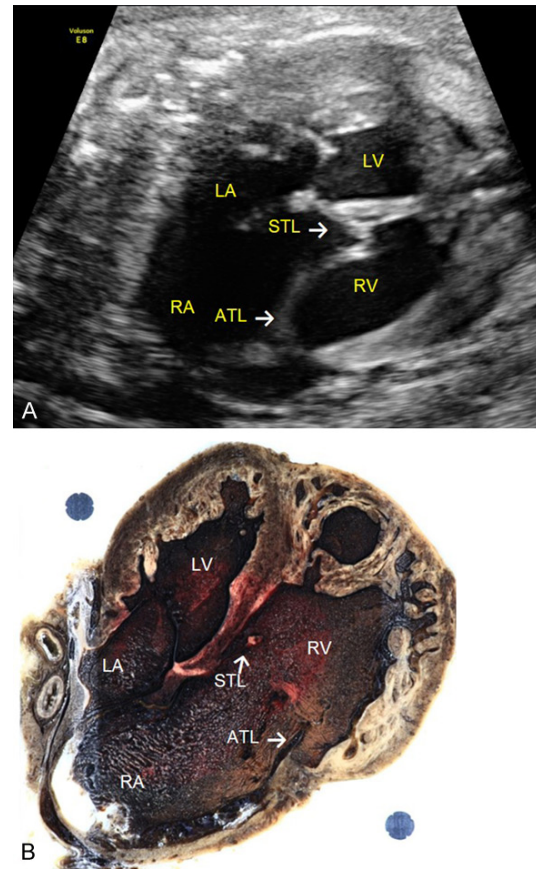


**Figure 5.** Ebstein anomaly associated with valve thickening. A. Ultrasonic image. B. Anatomical image. Abbreviations: LA, left atrium; RA, right atrium; LV, left ventricle; RV, right ventricle; ATL, anterior tricuspid leaflet; STL, septal tricuspid leaflet; PTL, posterior tricuspid leaflet; SP, spine.

leaflets were directly attached to the ventricular septum and the ventricular wall because of the absent chordae tendineae and papillary muscle. The septal and posterior tricuspid leaflets resembled a layer of fibrous tissue (**Figure 4**), consistent with the adult type.

#### *Ebstein anomaly associated with valve thickening*

A variety of valve abnormalities are associated with Ebstein anomaly. The majority of the downward-displacing leaflets are shortened, defective and adherent to the ventricular wall



**Figure 6.** Ebstein anomaly associated with ventricular noncompaction. A. Ultrasonic image. B. Anatomical image. Abbreviations: LA, left atrium; RA, right atrium; LV, left ventricle; RV, right ventricle; ATL, anterior tricuspid leaflet; STL, septal tricuspid leaflet.

[7]. Ebstein anomaly associated with valve thickening was rarely observed in the present study, and only a single case has previously been reported [8]. The observed case, constructed into a three-dimensional anatomical database after artificial labor [9], showed downward displacement of the septal and posterior tricuspid leaflets, with thickening and nodosity (**Figure 5**).

#### *Ebstein anomaly associated with ventricular noncompaction*

In recent years, left ventricular noncompaction (LVNC) is a disease that has been increasingly diagnosed in clinical practice [10]. And the relationship between Ebstein anomaly and LVNC has received increasing attention. The identification of LVNC in Ebstein anomaly has prognostic implications because this condition might be associated with left ventricular systolic dys-

function. LVNC can be an isolated finding or associated with other congenital cardiopathies, such as complex cyanotic heart diseases and, particularly, Ebstein anomaly [11]. Recent studies have identified mutations in the MYH7 gene linking Ebstein anomaly and LVNC [12]. The observed case, constructed into a three-dimensional anatomical database [9] after artificial labor, showed downward displacement of the septal tricuspid leaflet, a long anterior tricuspid leaflet, and enlargement of the right atrium, multiple prominent myocardial trabeculations and deep intratrabecular recesses in the ventricular wall (**Figure 6**).

In summary, Ebstein anomaly has a high mortality. Early diagnosis is beneficial to reduce birth defects. Fetal echocardiography has been the first choice for diagnosing Ebstein anomaly because this technique is not only accurate, rapid, simple, and inexpensive but also has a high diagnostic value. Although the ultrasonic appearance of typical Ebstein anomaly is becoming increasingly familiar, the rare type should also be considered, thereby increasing the current knowledge and providing additional clinical information to improve the diagnostic rate of Ebstein anomaly using ultrasound techniques.

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### Disclosure of conflict of interest

None.

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