Case Report
Obstet Gynecol Sci 2014;57(6):530-533
http://dx.doi.org/10.5468/ogs.2014.57.6.530
pISSN 2287-8572 · eISSN 2287-8580

Obstetrics & Gynecology Science

An unusual ultrasonographic manifestation of a fetal Ebstein anomaly

Min-Young Cha, Hye-Sung Won, Mi-Young Lee, Kyung-Hee Woo, Jae-Yoon Shim

Department of Obstetrics and Gynecology, University of Ulsan College of Medicine, Asan Medical Center, Seoul, Korea

An Ebstein anomaly is a rare congenital heart defect defined by an inferior displacement of the septal and posterior leaflets of the tricuspid valve from the tricuspid annulus. This anomaly shows various ultrasonographic manifestations, thus making the prenatal diagnosis sometimes difficult. We here report a rare case of an Ebstein anomaly which was prenatally suspected as the absence of the tricuspid valve with functional pulmonary atresia because of non-visible tricuspid leaflets on an echocardiograph at 24 weeks of gestation. An emergency cesarean section was performed at 35 weeks of gestation as fetal hydrops were seen on a follow-up scan. Postnatal surgery confirmed Ebstein anomaly type-D which demonstrates an almost complete atrialization of the right ventricle with the exception of a small and infundibular component. Because of its rarity, prenatal findings of a type-D Ebstein anomaly have not been reported previously. We suggest from this first such case report that this anomaly should be considered as a possible diagnosis when the tricuspid leaflets are not well visualized.

Keywords: Ebstein anomaly; Echocardiography; Fetal heart; Prenatal diagnosis

Introduction

Ebstein anomalies are rare congenital malformations occurring in about 1 to 5 per 200,000 live births, and accounting for less than 1% of all congenital heart diseases [1-3]. These anomalies are characterized by abnormalities of the tricuspid valve and right ventricle [4], which result a failed delamination of the leaflets from the interventricular septum, which leads to an adherence of these leaflets to the underlying myocardium [2,5,6]. Any degree of downward displacement of the tricuspid valve leaflets from the atrioventricular ring can be shown [3,4].

Fetal echocardiography is a useful tool for the diagnosis of congenital heart disease [2,4,7,8]. However as Ebstein anomalies show a wide spectrum of ultrasonographic manifestations, it can be difficult to make a prenatal diagnosis [2,3,5]. We here present a rare case of an Ebstein anomaly which demonstrated unusual ultrasonographic findings in the prenatal period, and discuss the diagnostic considerations in such patients.

Case report

A 29-year-old gravida 1, para 0 woman was referred to our tertiary center at 24.4 weeks of gestation with a suspected cardiac anomaly. Her previous medical history was unremarkable, and she had no family history of congenital malformation. Quad tests and other prenatal laboratory findings were within normal ranges. A fetal echocardiography was performed and the four-chamber view of the heart revealed cardiomegaly with a right ventricular dilatation without definite contractility, but no obvious right atrial dilatation (Fig. 1A). There were no discernible tricuspid leaflets (Fig. 1B), indicating the absence of a tricuspid valve structure. Color Doppler imaging showed forward and backward flows from the right atrium to the right ventricle without evidence of tricuspid regurgitation (Fig. 1C, D). Because the tricuspid valve structure

Received: 2014.5.30. Revised: 2014.7.16. Accepted: 2014.7.18.
Corresponding author: Hye-Sung Won
Department of Obstetrics and Gynecology, Asan Medical Center, 88 Olympic-ro 43-gil, Songpa-gu, Seoul 138-736, Korea
Tel: +82-2-3010-3644 Fax: +82-2-3010-6944
E-mail: hswon@amc.seoul.kr

was completely absent and also because there were communicating flows between right atrium and right ventricle, the fetus was thought to have a condition other than tricuspid atresia which is characterized with an absence of communication between right atrium and right ventricle. Functional pulmonary atresia was also associated. No other abnormalities were detected.

Following the counseling on the postnatal outcomes, the parents decided to continue the pregnancy, and did not want to undergo fetal karyotyping. Subsequent serial echocardiographic examinations showed no demonstrable changes. A follow-up evaluation at 35.5 weeks of gestation revealed newly developed skin edemas, pleural effusion, and ascites suggestive of fetal hydrops. An emergency cesarean section was therefore performed.

A male neonate of 2,160 g was delivered and was cyanotic with an Apgar score of 3 and 5 at 1 and 5 minutes, respectively, and an SaO_2 level of 50%. An initial echocardiography

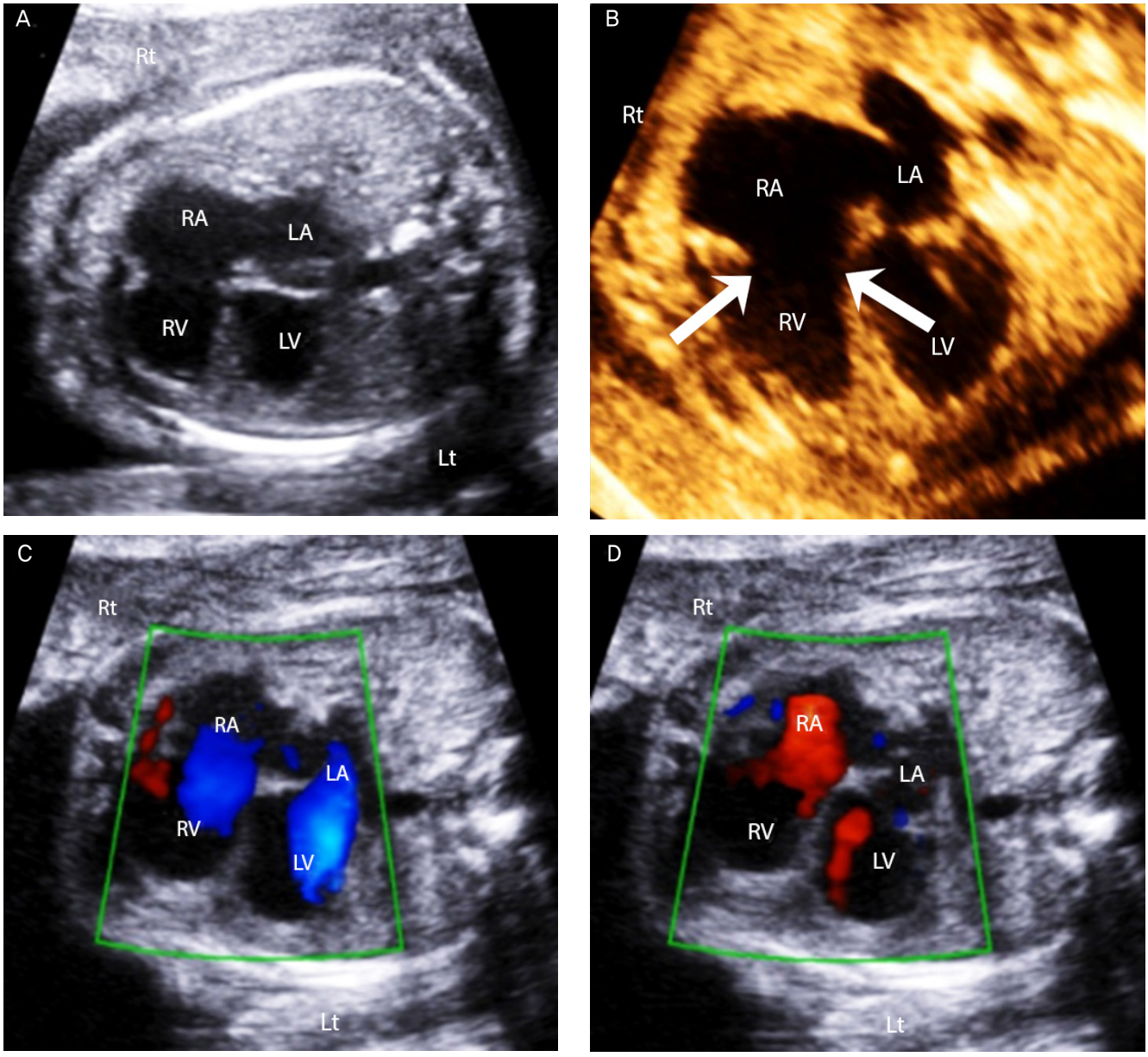

Fig. 1. Four-chamber view of the heart in the study patient showing the dilatation of the right ventricle, with no obvious dilatation of the right atrium (A). Three-dimensional ultrasound demonstrating no discernible tricuspid leaflets (arrows) (B). Color Doppler imaging showing a to-and-fro from the right atrium to the right ventricle (C,D). Rt, right; Lt, left; RA, right atrium; LA, left atrium; RV, right ventricle; LV, left ventricle.

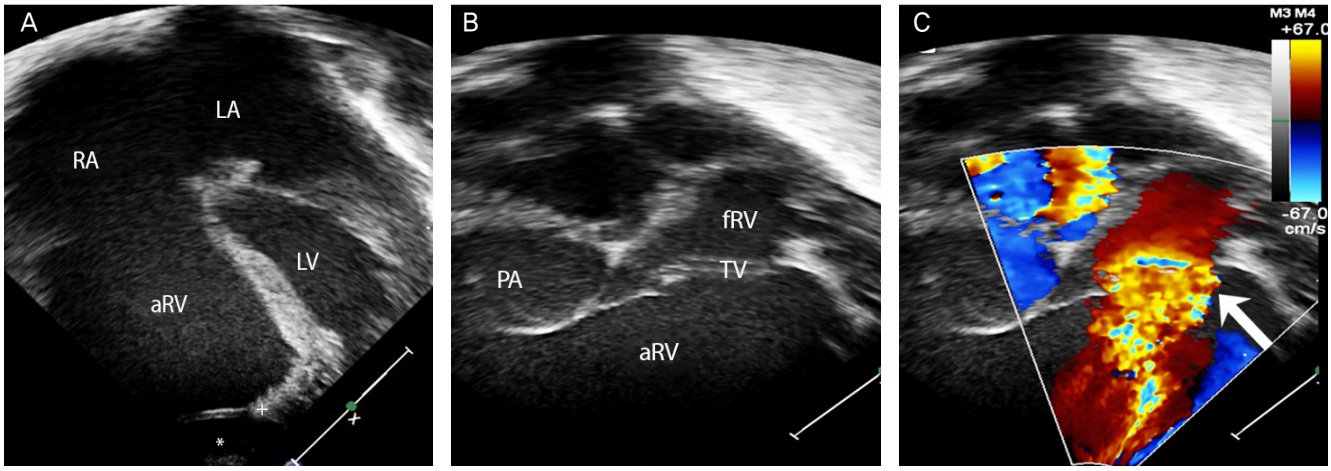

Fig. 2. Postnatal echocardiogram showing an almost completely atrialized right ventricle (aRV) with only a small outlet portion operating as a functional RV (*) and a severe downward displacement of the tricuspid valve leaflet insertion site (+) (A). Right ventricular outlet view also showing a very small functional RV (B), and severe tricuspid regurgitation on color Doppler (arrow) (C). RA, right atrium; LA, left atrium; LV, left ventricle; aRV, atrialized right ventricle; PA, pulmonary artery; fRV, functional right ventricle; TV, tricuspid valve.

indicated an Ebstein anomaly type-D, stenotic form with almost full atrialization of the right ventricle leaving only a very small functional portion of this ventricle (Fig. 2). A modified Starnes operation (right atrial reduction plasty with a right ventricular obliteration, central shunt and ligation of the patent ductus arteriosus) was performed on postnatal day 5, and surgical findings confirmed the initial diagnosis. Unfortunately the baby died on postoperative day 3 because of continuous mediastinal bleeding.

Discussion

To our knowledge, our current report is the first to describe the prenatal echocardiographic findings of a postnatally confirmed type-D Ebstein anomaly. A recent study from our center has described the management of 59 neonates with an Ebstein anomaly that had been treated in our division of pediatric cardiology between 2001 and 2012 [7]. In contrast to earlier reports that the majority of neonatal Ebstein anomalies are not diagnosed in the prenatal period [9,10], 46 of the patients (78%) at our center had been prenatally diagnosed with this condition [7]. Two babies in this series died immediately after birth, and among the remaining 57 neonates with an Ebstein anomaly, there were only 2 type-D cases (3.4%). The first of these type-D cases had been transferred from another institution after birth, and the other case had been prenatally diagnosed with an apparent apical displacement of the tricuspid leaflets, which was evident on a fetal echocardiogram. In our current case, the tricuspid valves were completely nonvisible and did not demonstrate the typical findings of Ebstein anomaly such as tricuspid regurgitation and right atrial enlargement in the prenatal period.

Targeted fetal echocardiography is a reliable technique for diagnosing Ebstein anomaly [2,4]. Typical features of this disease are recognized as cardiomegaly, apical displacement of the insertion of the tricuspid septal leaflet (>8 mm/m² body surface area) with a redundant elongated anterior tricuspid leaflet, right atrial enlargement which results from dilatation of the atrialized portion of the right ventricle, and marked tricuspid regurgitation [2-4]. Arrhythmia and fetal hydrops are also possible fetal manifestations of Ebstein anomaly [3]. Ebstein anomaly can be classified by its anatomical presentation [11]. Type A is designated as an adherence of the septal and posterior leaflets without a restriction of the functional right ventricle volume; type B means that the right ventricle has atrialized with a normal anterior leaflet; type C refers to cases with a stenotic anterior leaflet; and type D cases manifest with an almost complete atrialization of the ventricle, with the exception of a small infundibular component. Although the precise prevalence of each type of Ebstein anomaly has not been investigated, it is assumed that type D accounts for 3.5% to 8% of cases [7,12] and also that the prenatal diagnosis of type-D is rarer. In type-D cases, all of the leaflet tissue, even the anterior parts, is attached to the right ventricular wall. When the attachment of the tricuspid valve leaflets is

extremely distal, it is difficult to detect because it may be confused with the papillary muscle in the right ventricle [13]. In such cases, the right atrium and ventricle may mimic a single large sac, which results in a misdiagnosis if the examiners fail to identify the small infundibular ventricular portion. This was why our current cases had been misdiagnosed as an absent tricuspid valve rather than an Ebstein anomaly.

The prenatal diagnosis of an Ebstein anomaly will become more important in the future because the outcomes of surgical and conservative management have improved a great deal in the current era [4,7]. The prognosis for a fetus diagnosed with Ebstein anomaly has been extremely poor in the past with a reported mortality rate as high as 81% [9]. There have also been no clear indications in the past for surgical interventions in critically ill neonatal cases [3,4,9]. However with the gradual improvement in surgical technique and medical management, neonatal mortality has been reported at 7% to 24% and neonatal surgical mortality at a decreased rate of 23% to 27% [7,9,12,14].

In conclusion, Ebstein anomaly should be considered as a possible diagnosis when a tricuspid valve abnormality is suspected prenatally, even if there are no visible tricuspid leaflets. Awareness of several diagnostic pitfalls with careful ultrasonographic evaluation can help physicians to reach an accurate prenatal diagnosis in this regard. Furthermore, this will enable better and more informed prenatal counseling for parents and assist clinicians to adopt multidisciplinary approaches, including the induction of high-risk delivery, surgical intervention planning, and perinatal intensive care.

Conflict of interest

No potential conflict of interest relevant to this article was reported.

References

1. Dearani JA, Danielson GK. Congenital Heart Surgery Nomenclature and Database Project: Ebstein's anomaly and tricuspid valve disease. Ann Thorac Surg 2000;69:S106-17.
2. Attenhofer Jost CH, Connolly HM, Edwards WD, Hayes D, Warnes CA, Danielson GK. Ebstein's anomaly - review of a multifaceted congenital cardiac condition. Swiss Med Wkly 2005;135:269-81.
3. Paranon S, Acar P. Ebstein's anomaly of the tricuspid valve from fetus to adult: congenital heart disease. Heart 2008;94:237-43.
4. Melendres G, Ormsby EL, McGahan JP, Moon-Grady AJ, Towner D, Taylor D. Prenatal diagnosis of Ebstein anomaly: a potential pitfall. J Ultrasound Med 2004;23:551-5.
5. Krieger EV, Valente AM. Diagnosis and management of ebstein anomaly of the tricuspid valve. Curr Treat Options Cardiovasc Med 2012;14:594-607.
6. Anderson KR, Zuberbuhler JR, Anderson RH, Becker AE, Lie JT. Morphologic spectrum of Ebstein's anomaly of the heart: a review. Mayo Clin Proc 1979;54:174-80.
7. Yu JJ, Yun TJ, Won HS, Im YM, Lee BS, Kang SY, et al. Outcome of neonates with Ebstein's anomaly in the current era. Pediatr Cardiol 2013;34:1590-6.
8. Celermajer DS, Bull C, Till JA, Cullen S, Vassillikos VP, Sullivan ID, et al. Ebstein's anomaly: presentation and outcome from fetus to adult. J Am Coll Cardiol 1994;23:170-6.
9. Yetman AT, Freedom RM, McCrindle BW. Outcome in cyanotic neonates with Ebstein's anomaly. Am J Cardiol 1998;81:749-54.
10. Wald RM, Adatia I, Van Arsdell GS, Hornberger LK. Relation of limiting ductal patency to survival in neonatal Ebstein's anomaly. Am J Cardiol 2005;96:851-6.
11. Carpentier A, Chauvaud S, Mace L, Relland J, Mihaileanu S, Marino JP, et al. A new reconstructive operation for Ebstein's anomaly of the tricuspid valve. J Thorac Cardiovasc Surg 1988;96:92-101.
12. Chauvaud S. Ebstein's malformation: surgical treatment and results. Thorac Cardiovasc Surg 2000;48:220-3.
13. Tongsong T, Chanprapaph P, Khunamornpong S, Sirichotiyakul S. Sonographic features of Ebstein anomaly associated with hydrops fetalis: a report of two cases. J Clin Ultrasound 2005;33:149-53.
14. Davies RR, Pasquali SK, Jacobs ML, Jacobs JJ, Wallace AS, Pizarro C. Current spectrum of surgical procedures performed for Ebstein's malformation: an analysis of the Society of Thoracic Surgeons Congenital Heart Surgery Database. Ann Thorac Surg 2013;96:1703-9.