Undiagnosed Ebstein's Anomaly in a Pregnant Woman

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Ebstein's anomaly is an uncommon congenital heart defect characterized by an abnormal tricuspid valve. Pregnancy can be well tolerated, although cyanosis, arrhythmias, and cardiomegaly are associated with poor prognosis. An 18-year-old woman with a history of congenital cardiomyopathy presented at 37 weeks of gestation with hypoxia, dyspnea, central cyanosis, and uterine contractions. The patient underwent emergent cesarean delivery. Echocardiography revealed Ebstein's anomaly with patent foramen ovale and right-to-left shunting. Tricuspid valvuloplasty with primary closure of patent foramen ovale was performed. This case is presented, accompanied by a review of the literature.

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KEY WORDS

Ebstein's anomaly • Pregnancy • Congenital heart disease

Ebstein's anomaly is a rare congenital heart defect involving abnormalities of the tricuspid valve. Incidence is 1 per 200,000 live births, accounting for approximately 0.3% to 0.7% of all cases of congenital heart disease.[1,2] In patients with Ebstein's anomaly, pregnancy is usually well tolerated unless cyanosis or arrhythmia develops; yet there is an increased risk of prematurity and low birth weight.[3] Ebstein's anomaly can be accurately identified by echocardiography. Women with Ebstein's anomaly should be carefully assessed and followed by a multidisciplinary team.

Case Report

An 18-year-old white woman gravida 2 para 0 abortion 1 diagnosed with asthma and a congenital heart disease at birth presented at 37 weeks of gestation with uterine contractions. She had no regular antenatal care and remained well despite chronic hypoxia and cyanosis at baseline until admission, when she had nausea, vomiting, and dyspnea. She denied chest pain, lightheadedness, syncope, or palpitations.

Upon arrival, her blood pressure was 113/75 mm Hg. Heart rate was 77 beats/min and regular with a respiratory rate of 24 breaths/min. Physical

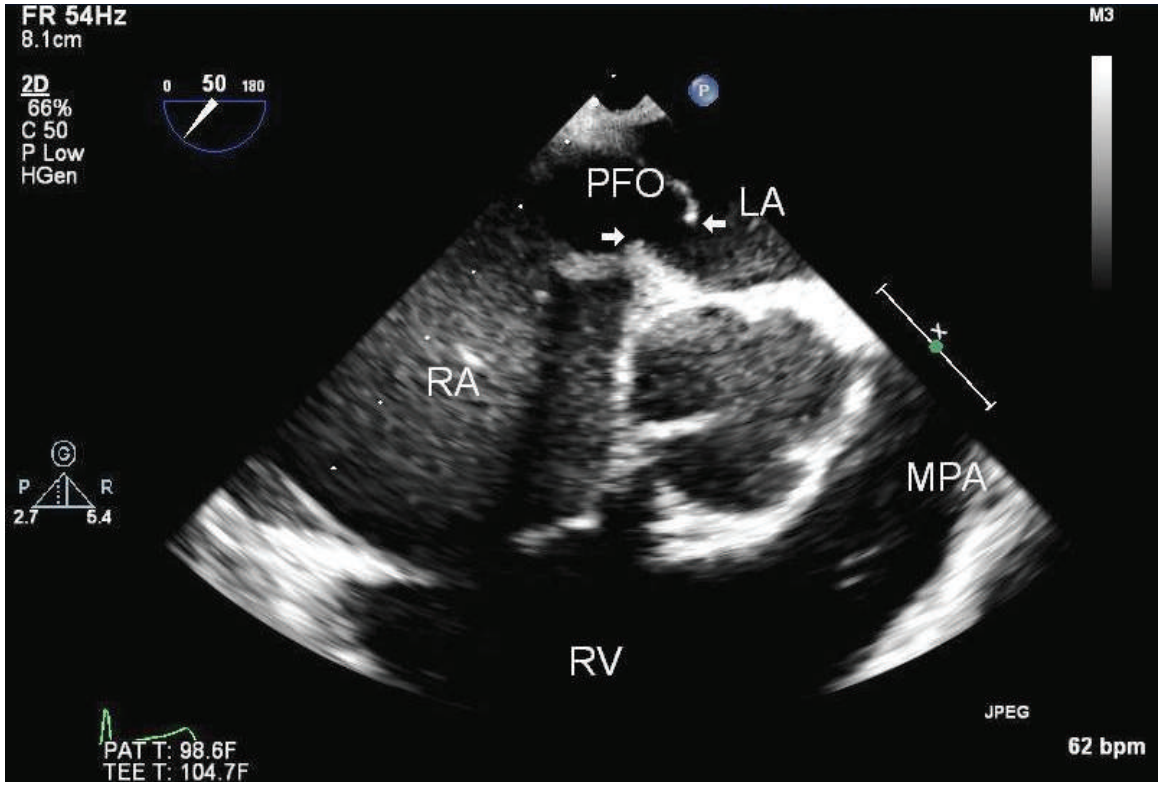
examination revealed central cyanosis. On cardiac auscultation, a grade 2 blowing systolic murmur was heard in the tricuspid area and lung fields were clear. In arterial blood gas analysis, her arterial oxygen tension was 58 mm Hg (saturation 82%) breathing on room air, which improved to 88 mm Hg (saturation 94%) on 15 L/min oxygen therapy.

Blood tests showed a hemoglobin level of 13.1 g/dL (hematocrit at 38.2%) and a mild leukocytosis at 15.7 $\times$ 10^3 μL; otherwise, complete blood count, coagulation, and biochemistry results were within normal limits.

An electrocardiogram showed sinus rhythm at 77 beats/min with premature atrial complexes and an incomplete right bundle branch block (RBBB). Chest radiography showed cardiomegaly. Chest computed tomography did not reveal evidence of interstitial lung disease or pulmonary embolus. On the day of admission, the patient underwent cesarean delivery under general anesthesia due to progressive desaturation despite oxygenation therapy and history of cardiomyopathy. The surgery was uneventful. A live infant girl weighing 2610 g was delivered with Apgar scores of 8 at 1 minute and 9 at 5 minutes. Transesophageal echocardiogram after cesarean delivery showed inferior displacement of a moderately dilated right atrium with markedly dilated atrialization of the right ventricle, severe tricuspid incompetence, and a patent foramen ovale (PFO) with right-to-left shunting (Figure 1). Cyanosis and hypoxia persisted throughout the postoperative period. A consult was obtained with the pediatric cardiac surgeon and the patient was accepted for surgical correction of Ebstein's anomaly. Patient was discharged 5 days later.

Eight weeks postpartum, the patient underwent left and right heart cardiac catheterization, showing significant desaturation in the right atrium (55%) with pressure of 10 mm Hg, the right ventricle (53%) with pressure of 13/6 mm Hg, and the pulmonary artery (52%) with pressure of 15/9 mm Hg (indicating no pulmonary hypertension); left atrial pressure was 4 mm Hg, left ventricular pressure was 114/4 mm Hg, and aortic pressure was 124/75 mm Hg. A left ventricular angiogram revealed an ejection fraction of 45%, and a hypokinetic right ventricle. Repeat echocardiography revealed (1) apical displacement of the septal tricuspid valve leaflet by 9 mm, (2) adherence of the septal leaflet to the myocardium, (3) moderate dilation of the right atrium, (4) dilation of the atrialized portion of the right ventricle with global hypokinesis, (5) stretch PFO with bidirectional atrial level

Figure 1. Transesophageal echocardiogram showing a PFO (*arrows*). It shows a grossly enlarged RA, along with dilation of the atrialized RV. LA , left ventricle; MPA, main pulmonary artery; PFO, patent foramen ovale; RA, right atrium; RV, right ventricle.



shunting, and (6) severe tricuspid regurgitation.

The patient underwent primary closure of the PFO as well as tricuspid valvuloplasty using a 28-mm Carpentier-Edwards ring (Edwards Lifesciences, Irvine, CA). Surgery was uneventful and resulted in improvement in oxygenation saturation, ranging from 89% to 94% on room air (compared with preoperative saturations ranging from 72% to 84% on room air). The patient was discharged on postoperative day 5 in good hemodynamic condition with oxygen saturations > 92% on room air. She was doing well without any symptoms on follow-up.

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Discussion

Ebstein's anomaly is an uncommon congenital heart defect with a prevalence of less than 1% without any predilection for either sex.[1,2] The survival of some patients with Ebstein's anomaly during childhood and adolescence has improved drastically, and patients have been reported to live up to 85 years.[4] Ebstein first described the aberration of the tricuspid valve in 1866. It is characterized by an abnormal septal tricuspid leaflet that is displaced apically from the atrioventricular ring into the right ventricle. It has a distal effective portion and an atrialized proximal portion.[5] Although other types of tricuspid valve disease (rheumatic, endocarditic, connective, traumatic, carcinoid, dysplastic) can mimic Ebstein's anomaly,[6] apical displacement of the septal leaflet of the tricuspid valve from the insertion of the anterior leaflet of the mitral valve by at least 8 mm/m² is the principal feature of Ebstein's anomaly.[7]

An intratrial connection is present in 80% to 94% of patients with Ebstein's anomaly.[8,9] In the presence of an interatrial communication, the risk of paradoxical embolization, brain abscess, and sudden death increases.[10] Our patient had a PFO with a right-to-left shunt. Accessory pathways, most commonly Wolff-Parkinson-White syndrome, are not an uncommon finding in patients with Ebstein's anomaly (6%-26%).[11] Primary atrioventricular block is found in up to 50% of patients with Ebstein's anomaly; RBBB is also a typical finding.[4,12] Our patient did not have any signs of pre-excitation; electrocardiogram revealed an incomplete RBBB.

Women with Ebstein's anomaly without cyanosis and heart failure are classified into World Health Organization risk class II and tolerate pregnancy fairly well. The hemodynamic problems seen during pregnancy depend largely on the severity of the tricuspid regurgitation and the functional capacity of the right ventricle. In contrast, symptomatic patients with cyanosis and/or heart failure should be treated before pregnancy or counseled against pregnancy.[13] The degree of maternal hypoxemia is the most important predictor of fetal outcome. If resting oxygen saturation is < 85%, a substantial maternal and fetal mortality risk is expected and pregnancy is contraindicated; chances of a live birth are 12% as compared with 92% for women with saturations > 90%.[14] Oxygen therapy may slightly improve maternal saturation by pulmonary vasodilation and increased blood flow, but there is no evidence that this improves either maternal or fetal outcome.[15] Our patient had a history of low oxygen saturation and persistent cyanosis. Throughout her hospital stay, her resting oxygen saturations ranged 72% to 84% on room air and 82% to 96% on oxygenation therapy.

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Many important physiologic changes occur within the cardiovascular system during pregnancy. The plasma volume increases by 40% to 50%, whereas erythrocyte volume increases by only 15% to 20%, which causes a situation that is described as physiologic anemia of pregnancy. Cardiac output rises by 40%, with an increase in both stroke volume and heart rate. Systemic blood pressure is lower than in nonpregnant women, indicating a considerable reduction in peripheral vascular resistance and placental shunting.[3] These changes may aggravate the hemodynamic consequences in patients with Ebstein's anomaly, resulting in worsening tricuspid regurgitation, precipitation of heart failure, raised atrial pressures, and increased right-to-left shunting.[16] Severe exertion and possibly anesthesia during labor may produce sudden death.[17] No correlation was demonstrated between symptoms and the degree of displacement of the tricuspid valve leaflets.[18]

The presence of arrhythmia or cyanosis in the mother is associated with increased maternal and fetal risk, and requires closer maternal and fetal observation during pregnancy and delivery.[3] Maternal complications (heart failure, pulmonary or systemic thrombosis, supra-

ventricular arrhythmias, infective endocarditis) occur in 30% of cyanotic pregnant patients.[13] Mothers with cyanosis are at increased risk of miscarriage/fetal loss, premature delivery, and infants with low birth weight.[3,19] Connolly and Warnes[19] studied 111 pregnancies in 44 women with Ebstein's anomaly. The mean birth weight of infants born to cyanotic women was significantly lower than that of newborns of acyanotic women (2530 vs 3140 g; $P < 0.001$).[19] The babies had a birth weight of 2160 g and were diagnosed with intrauterine fetal growth retardation due to the combined effects of maternal hypoxemia and continued use of tobacco, alcohol, and cannabis throughout pregnancy. In general, the perinatal outcome was favorable. The preferred mode of delivery is vaginal in almost all cases unless maternal or fetal condition deteriorates.[13] There is a 4% to 6% risk of congenital heart disease in offspring and a 0.6% risk of familial Ebstein's anomaly.[3,20] Our patient underwent emergent cesarean delivery due to progressive hypoxia despite oxygenation therapy and history of congenital heart defect.

Possible prognostic factors in Ebstein's anomaly have been assessed in several studies and those features associated with poor outcome include (1) other complicating cardiac lesions, (2) persistent cyanosis as a result of right-to-left atrial shunting, (3) diagnosis made in infancy, (4) severe cardiomegaly, (5) development of dyspnea at rest, and (6) right heart failure.[4,21-23] Our patient survived her pregnancy despite having three of the poor prognostic features (PFO, cardiomegaly, and persistent cyanosis secondary to right-to-left shunting).

The mean birth weight of infants born to cyanotic women was significantly lower than that of newborns of acyanotic women.

This case highlights the variable anatomic and clinical spectrum of Ebstein's anomaly and its effects on pregnancy. This case is unique in that a tricuspid annuloplasty was able to reduce Ebstein's anomaly-related tricuspid regurgitation with no complications at follow-up. It also confirms the importance of echocardiographic evaluation and a multidisciplinary approach for successful outcome in high-risk patients. ■

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MAIN POINTS

- Ebstein's anomaly is a rare congenital heart defect involving abnormalities of the tricuspid valve and accounts for approximately 0.3% to 0.7% of all cases of congenital heart disease.
- Pregnancy is usually well tolerated in patients with Ebstein's anomaly, unless cyanosis or arrhythmia develops. There is an increased risk of prematurity and low birth weight. Hemodynamic problems seen during pregnancy depend largely on the severity of the tricuspid regurgitation and the functional capacity of the right ventricle.
- Symptomatic patients with cyanosis and/or heart failure should be treated before pregnancy or counseled against pregnancy.
- Echocardiographic evaluation and a multidisciplinary approach are required for successful outcome in high-risk patients.

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