

Late-Onset Cyanosis and Finger Clubbing in a 37-Year-Old Female with Ebstein's Anomaly and Atrial Septal Defect

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It is common to suspect Eisenmenger syndrome when patients with atrial septal defect (ASD) show cyanosis and clubbing. However, ASD accompanying Ebstein's anomaly frequently shows cyanosis without severe pulmonary hypertension, especially in the early period after birth. So the medical decision to decline surgical treatment in those patients should not be determined merely by clinical features such as cyanosis and clubbing. We report a case of a 37-year-old female with Ebstein's anomaly and secundum ASD who showed lip cyanosis and finger clubbing, but maintained normal pulmonary artery pressure and thus underwent successful surgical correction.

Key Words: Atrial septal defect • Cyanosis • Ebstein's anomaly • Eisenmenger syndrome

INTRODUCTION

Less than 1% of patients with atrial septal defect (ASD) develop severe pulmonary hypertension with shunt reversal, the so-called ASD-Eisenmenger syndrome.¹ Although practitioners usually suspect Eisenmenger syndrome when patients with ASD show cyanosis and clubbing, some of them can show cyanosis without severe pulmonary hypertension.² We report a case of a 37-year-old female with Ebstein's anomaly and ASD who manifested lip cyanosis and finger clubbing, but, maintained normal pulmonary artery pressure and thus underwent successful surgical correction.

CASE REPORT

A 37-year-old female who had undergone surgical ASD closure 26 years ago presented with progressive dyspnea during the past 10 years. She complained of only chest discomfort before her previous surgery. Three years ago, she was diagnosed as Eisenmenger syndrome in another hospital, solely on the basis of her medical history and physical findings of cyanosis and finger clubbing; these characteristics were not evident 10 years ago, and thus she remained in an inoperable state without further evaluation. On arrival, she showed lip cyanosis, finger clubbing and neck vein engorgement with stable vital signs (blood pressure 100/60 mmHg, pulse rate 62 beats/minute, respiration rate 20/minute and body temperature 36.4 °C). Auscultation revealed grade III systolic murmur over the apex of the heart. Electrocardiography (Figure 1A) showed atrial fibrillation and right bundle branch block. A chest X-ray of the patient showed marked cardiomegaly with diminished pulmonary vascularity and a trace of median sternotomy (Figure 1B). Laboratory testing revealed erythrocytosis (hemoglobin 17.5 g/dL), hypoxemia (SpO₂ 85% in room air) and jaundice (total bilirubin 4.4

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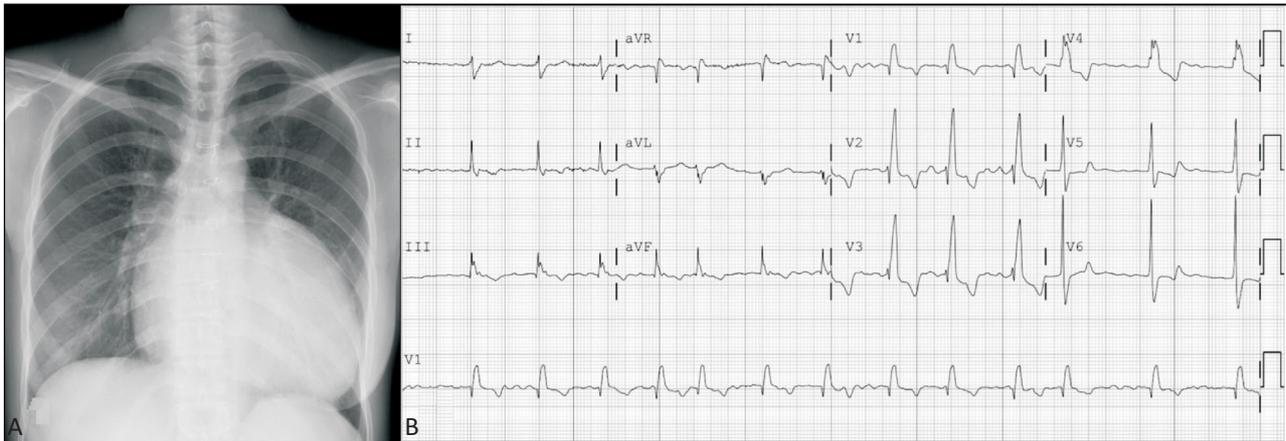


Figure 1. (A) Chest X-ray showing marked cardiomegaly with diminished pulmonary vascularity. (B) Electrocardiography showing atrial fibrillation and right bundle branch block.

mg/dl). Transthoracic echocardiography revealed apical displacement of the septal leaflet of the tricuspid valve (TV), coaptation failure of TV, dilatation of right-sided chambers and a 6 mm-sized tissue defect in the mid-portion of the interatrial septum compatible with secundum ASD (Figure 2A). There was evidence of right ventricular (RV) hypertrophy (thickness of RV free wall 6 mm) and decreased RV systolic function (peak systolic velocity of RV free wall by pulsed wave tissue Doppler imaging 9.7 cm/s, RV ejection fraction by Simpson's method 43%). Color flow imaging and pulsed wave Doppler tracing revealed left to right shunt (L-R shunt) during diastole and right to left shunt (R-L shunt) during systole when severe tricuspid regurgitation (TR) occurred (Figure 2B). The estimated peak systolic pressure gradient between the patient's right atrium (RA) and right ventricle was only 12.8 mmHg (Figure 2C). Furthermore, her pulmonary artery was not dilated.

Right heart catheterization was performed to assess further hemodynamic parameters. The corresponding right heart pressure consisted of a mean right atrial pressure of 17 mmHg, RV end-diastolic pressure of 16 mmHg, and mean pulmonary arterial pressure of 20 mmHg (Figure 2D). The result of oximetric measurements were compatible with bidirectional shunt as shown in echocardiography (inferior vena cava: 61.4%; superior vena cava: 59.2%; upper RA: 70.0%; mid RA: 70.7%; lower RA: 70.0%; and aorta: 84.5%). The estimated ratio of pulmonary blood flow to systemic blood flow (Q_p/Q_s) was 0.88. Additionally, the calculated pulmonary vascular resistance was 1.5 Wood units.

In light of these results, we concluded that the patient was a satisfactory surgical candidate. The actual procedure consisted of primary closure of ASD, tricuspid valve replacement with a 31-mm Perimount Magna bioprosthesis (Edwards Lifesciences, Irvine, CA, USA) and right atrial maze procedure with cryoablation. To relieve the RV volume overload, superior vena cava was anastomosed to the right pulmonary artery using end-to-side anastomosis technique. The patient complained of swelling of her face and upper arm during the immediate postoperative period, which gradually subsided with medical treatment including diuretics. She also suffered from transient right ventricular failure and pneumonia, but was eventually discharged without dyspnea 47 days after surgery.

DISCUSSION

It is well-known that patients with unrepaired ASD rarely develop Eisenmenger syndrome; however, 10% of patients with large unrepaired ASD are at risk of developing Eisenmenger syndrome.^{1,3} If adult ASD patients show cyanosis, Eisenmenger syndrome should be considered. Kai et al. showed that R-L shunt could occur without severe pulmonary hypertension and TR was the major determinant in their study of 36 ASD patients with transesophageal echocardiography.⁴ Oki et al. analyzed 26 patients who had secundum type ASD with R-L shunt. They reported that 5 patients who had elevated mean RA pressure without pulmonary hyper-

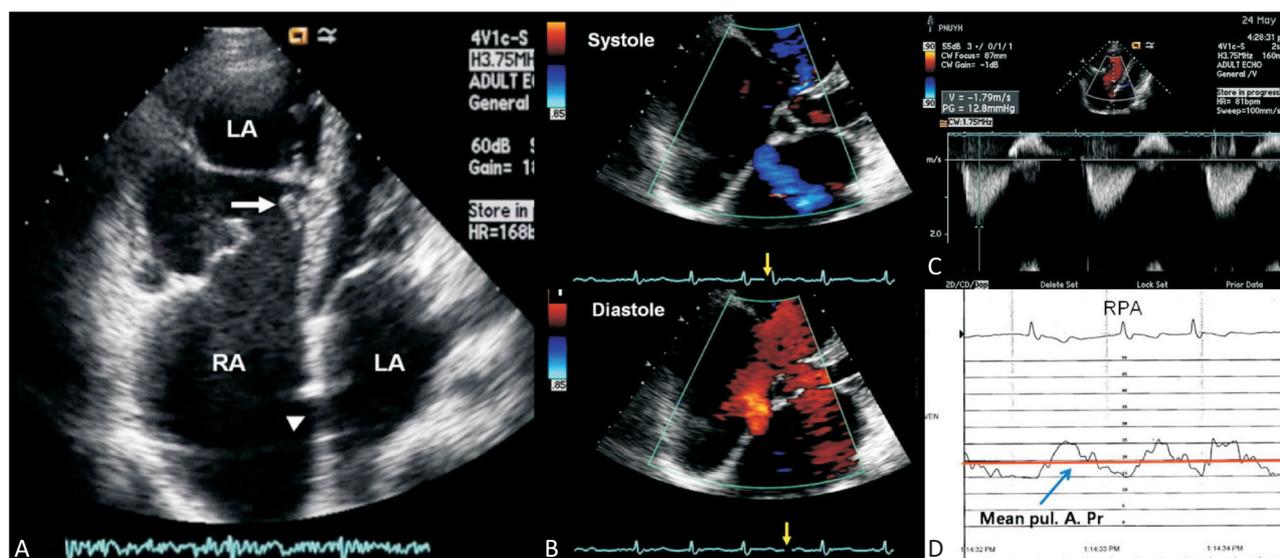


Figure 2. (A) Apical four chamber view of transthoracic echocardiography demonstrating apical displacement of the septal leaflet of tricuspid valve (arrow), dilatation of both right atrium and ventricle, coaptation failure of tricuspid valve and 6 mm-sized defect of interatrial septum (arrow head). (B) Color flow image showing right to left shunt during systole when severe tricuspid regurgitation occurs and left to right shunt during diastole. (C) Estimated peak pressure gradient between right atrium and right ventricle was only 12.8 mmHg. (D) Cardiac catheterization showing mean pulmonary artery pressure of 20 mmHg.

tension showed R-L shunt flow at the onset of ventricular contraction or the closing phase of TV; 6 patients who had TR without pulmonary hypertension showed R-L shunt flow during ventricular systole; and 5 patients who had severe pulmonary hypertension or pulmonic stenosis showed R-L shunt flow during atrial systole.⁵

About 80% of patients with Ebstein's anomaly have an interatrial communication (atrial septal defect or patent foramen ovale) through which right-to-left shunting of blood may occur.⁶ Cyanosis can be seen in the first days of life. Transient improvement may occur as pulmonary vascular resistance falls, but the condition worsens after the ductus arteriosus closes, thereby decreasing pulmonary blood flow. Jaiswal et al. reported that cyanosis was present in 48% and clubbing in 41% of patients with Ebstein's anomaly.⁷

In our case, the patient's medical records indicated that cyanosis and clubbing was not evident 10 years ago. It is presumed that a delayed onset of cyanosis and clubbing in our patient could be attributed to a gradual increase in RA pressure and worsening of TR severity with the passing of time. During right ventricular systole, large amount of regurgitant blood flow produced by TR might have raised RA pressure and eventually

reversed pressure gradient between RA and LA and thus made dominant R-L shunt flow and arterial hypoxemia causing cyanosis without pulmonary hypertension even in resting state. Therefore we thought that Ebstein's anomaly and severe TR was the cause of R-L shunt flow in this case. This speculation was also supported by the Qp/Qs. Although the shunt flow via ASD was bidirectional, estimated Qp/Qs of 0.88 meant that R-L shunt flow was predominant and thus could make our patient develop significant hypoxemia. The determinant of survival were pulmonary blood flow, cyanosis, clubbing, and systemic arterial oxygen saturation.⁷ Repair or replacement of the tricuspid valve in conjunction with closure of the interatrial communication is recommended for older patients who have severe symptoms despite medical therapy. In cyanotic patients with significant right-to-left shunting, the bidirectional cavopulmonary shunt reduces the volume load on the dysfunctional RV which permits safe closure of the intra-atrial communication in these patients.⁸

In addition, careful interpretation is needed when pulmonary arterial pressure is estimated by echocardiography. Fisher et al. reported that the pulmonary arterial pressure estimated by Doppler echocardiography was at least 10 mmHg higher or lower than that ob-

tained by right heart catheterization in 48 percent of 65 patients with various type pulmonary hypertension. Over- and under-estimation of pulmonary arterial pressure occurred with similar frequency, although the magnitude of the underestimation was greater.⁹

In conclusion, even in patient with cyanosis and long history of ASD, careful approach using multiple diagnostic modalities would be needed to exclude Eisenmenger syndrome and to determine the operability precisely.

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