A case of severe Ebstein's anomaly with incompetent pulmonary valve

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A case of Ebstein's anomaly with functional pulmonary atresia diagnosed in utero is presented. The diagnosis was confirmed by postnatal echocardiographic, angiographic, and postmortem pathologic findings. On echocardiography the septal leaflet of the tricuspid valve was displaced towards the right ventricular apex. The tricuspid valve was moderately regurgitant and the arterial duct was patent. Continuous wave or color Doppler revealed serious reduction in forward flow from right ventricle through the pulmonary arteries; however, massive pulmonary regurgitation was observed. Pulmonary circulation was dependent on the ductal flow due to functional pulmonary atresia. Angiography revealed the massively enlarged right atrium, the absence of forward flow through the tricuspid valve, transfer of contrast material through the atrial septal defect to the left atrium, and the retrograde inflow of the pulmonary arteries from the aorta via the patent arterial duct.

Ebstein's anomaly accompanied by functional pulmonary atresia is very rare. The fetal and neonatal presentation of this anomaly is associated with poor outcome.

Key words: Ebstein's anomaly, neonate, functional pulmonary atresia.

Ebstein's anomaly of the tricuspid valve is an uncommon malformation representing 0.3%-0.6% of all cases of congenital heart disease[1-3]. The basic defect involves a deformed and apically displaced tricuspid valve, resulting in symptoms that vary with age of the patient and the severity of the defect[1,4]. However, high pulmonary vascular resistance in the neonatal period may aggravate tricuspid regurgitation and lead to functional pulmonary atresia. Infants with severe Ebstein's anomaly and functional pulmonary atresia remain dependent on ductal patency. Hence this anomaly carries a high mortality rate in the neonatal period[1,2].

The purpose of this report is to describe and to discuss a neonate with severe Ebstein's anomaly, accompanied by functional pulmonary atresia. This iss a rare case, in which clinical, hemodynamic, angiographic, and pathological data are available.

Case Report

The mother of the patient was referred to the Pediatric Cardiology Unit in the 19th week of pregnancy with the suspicion of Ebstein's anomaly detected at routine fetal ultrasonographic evaluation. Fetal echocardiography revealed severe Ebstein's anomaly of the fetus.

The baby was delivered at the 35th week of gestation due to oligohydramnios and reduced fetal movements. The Apgar scores at 5 and 10 minutes postnatally were determined as 3 and 5, respectively. The baby was resuscitated because of absence of spontaneous respiration, severe cyanosis and bradycardia. Immediate postnatal echocardiographic study revealed displacement of the septal leaflet of the tricuspid valve towards the right ventricular apex. The distane between the tricuspid valve annulus and ventricular insertion of the septal leaflet was 11.8 mm (Fig. 1). The tricuspid valve was moderately regurgitant and the arterial duct was patent. Continuous wave or color Doppler revealed serious reduction in forward flow from right ventricle through the pulmonary arteries; however, massive pulmonary regurgitation was observed (Fig. 2). Pulmonary circulation was dependent on the ductal flow due to functional pulmonary atresia.

Cardiac catheterization via umbilical vein was performed; however, the catheter could not be advanced through the tricuspid valve to the right

ventricle and the pulmonary artery. Instead, it was advanced through the atrial septal defect to the left atrium and left ventricle. Right atrial and left ventricular angiograms were obtained. The right atrial angiogram (Figs. 3 and 3a) revealed the massively enlarged right atrium and the absence of forward flow through the tricuspid valve. All the contrast material passed through the atrial septal defect to the left atrium. The left ventriculogram (Fig. 4, 4a, 5 and 5a) demonstrated the retrograde inflow of the pulmonary arteries from the aorta via the patent arterial duct. Due to severe pulmonary regurgitation, contrast material reached the right ventricle and subsequently the right atrium via the regurgitant apically displaced tricuspid valve.

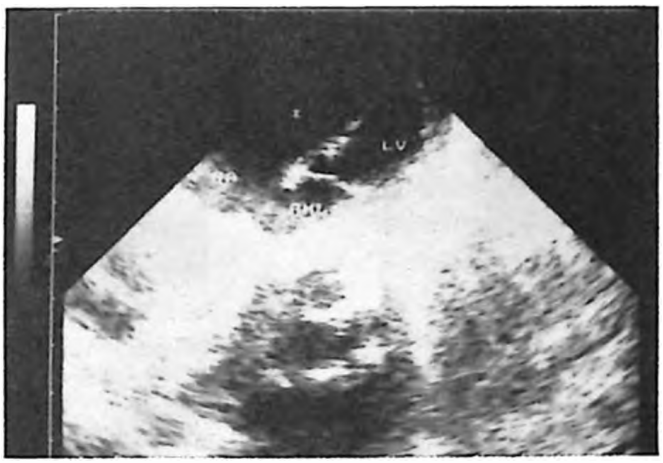

Fig. 1. The four-chamber view showing the mitral valve attachment. The right atrium is enlarged, and the tricuspid valve is greatly displaced towards the right ventricle and cannot be seen. The asterisk indicates the atrialized right ventricle. RA: right atrium, LV: left ventricle.

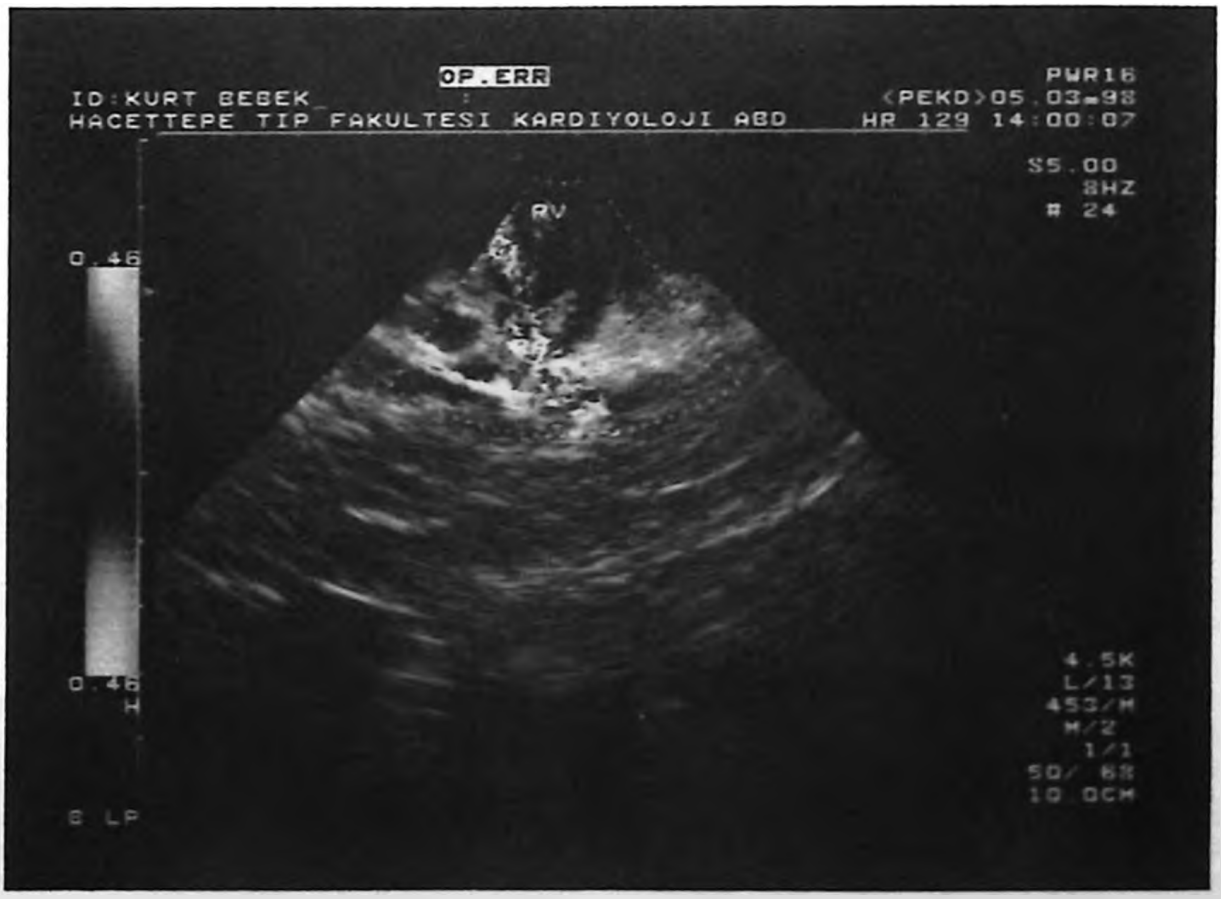

Fig. 2. A modified parasternal short-axis view showing severe pulmonary valve regurgitation. RV: right ventricle, PA: pullmonary artery.

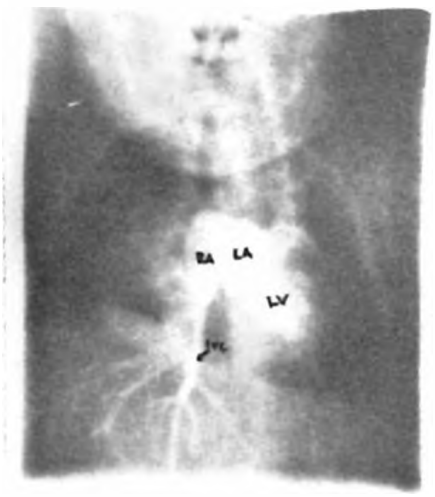

Fig. 3. The right atrial injection at antero-posterior position. Contrast material is seen in the right atrium. It travels to the left atrium and the left ventricle via the atrial septal defect. IVC: inferior vena cava, RA: right atrium, LA: left atrium, LV: left ventricle.

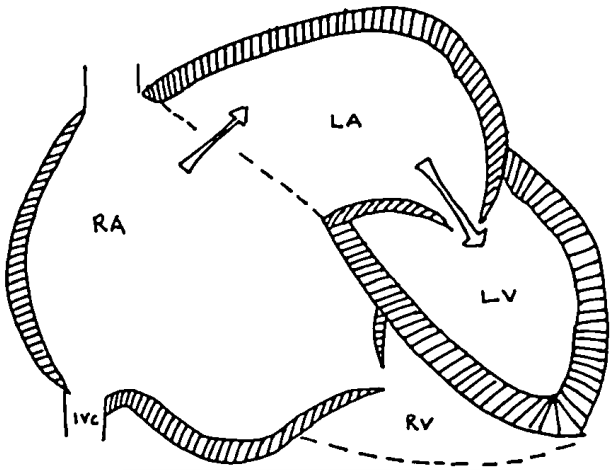

Fig. 3a: The right atrial injection at antero-posterior position (illustration of Fig. 3). Contrast materil is seen in the right atrium. It travels to the left atrium and the left ventricle via the atrial septal defect. IVC: inferior vena cava, RA: right atrium, LA: left atrium, LV: left ventricle.

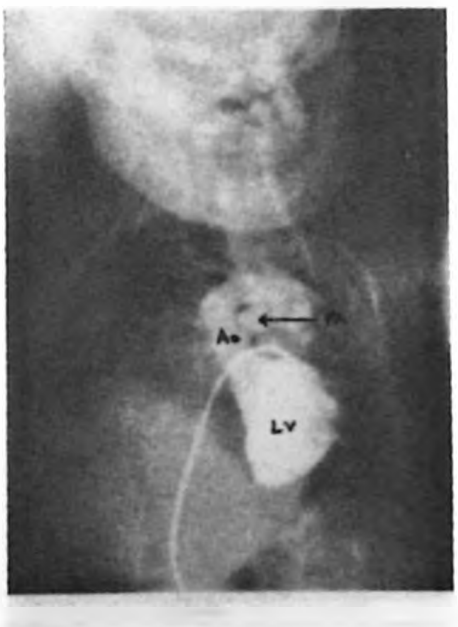

Fig. 4. The left ventricle injection in antero-posterior position (early phase). The left ventricle with intact ventricularseptum and the retrograde inflow of the pulmonary arteries from the aorta via the patent arterial duct are seen. LV: left ventricle, Ao: aorta, PA: pulmonary artery.

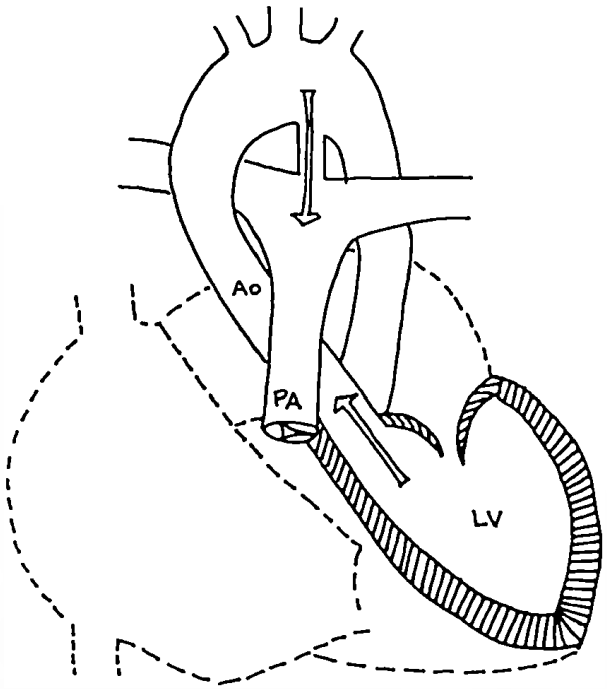

Fig. 4a. The left ventricle injection in antero-posteriorposition in early phase (illustration of Fig. 4). The left ventricle with intact ventricular septum and the retrograde inflow of the pulmonary arteries from the aorta via the patent arterial duct are seen. LV: left ventricle, Ao: aorta, PA: pulmonary artery.

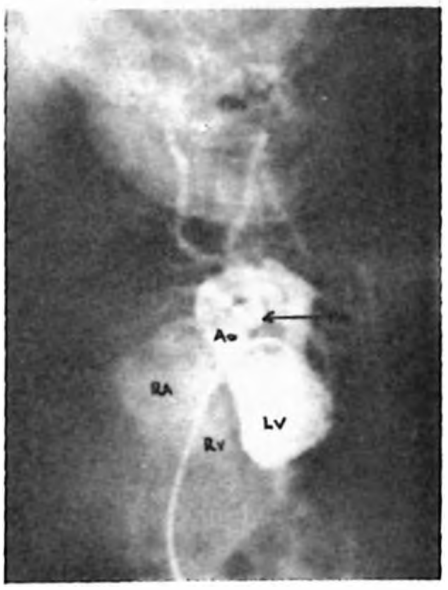

Fig. 5. The left ventricle injection in antero-posterior position (late phase). There is retrograde inflow to the pulmonary arteries from the aorta via the patent arterial duct. Due to severe pulmonary regurgitation, contrast material reaches the right ventricle and subsequently the right atrium via the regurgitant ricuspid valve. LV: left ventricle, Ao: aorta, PA: pulmonary artery, RV: right ventricle, RA: right atrium.

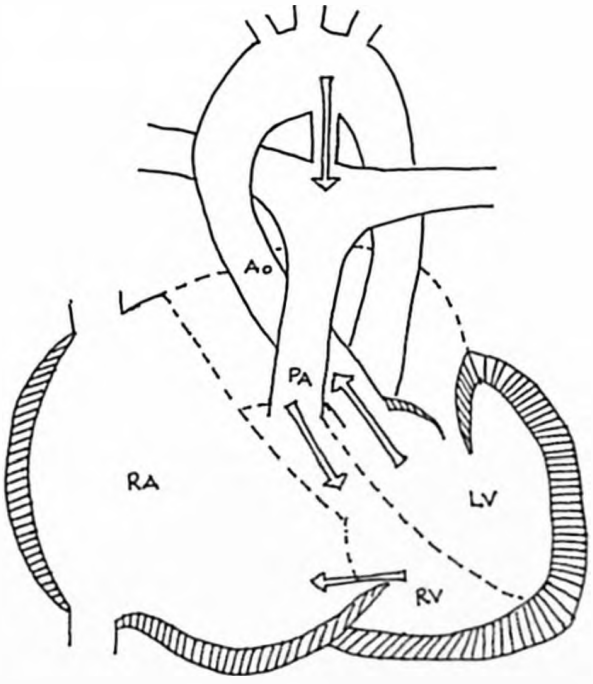

Fig. 5a. The left ventricle injection in antero-posterior position in late phase (illustration of Fig. 5). There is retrograde inflow to the pulmonary arteries from the aorta via the patent arterial duct. Due to severe pulmonary rergurgitation, contrast material reaches the right ventricle and subsequently the right atrium via the regurgitant ricuspid valve. LV: left ventricle, Ao: aorta, PA: pulmonary artery, RV: right ventricle, RA: right atrium.

The patient died of severe heart failure at the 9th hour of birth, despite aggressive resuscitation and prostaglandin E1 infusion. The postmortem examination revealed severe Ebstein's anomaly with a wide atrial septal defect, right atrial dilatation, a large patent arterial duct and a mild pulmonary valvular stenosis. ;Thickened and nodular leaflets were observed on the tricuspid and pulmonary valves. Microscopic examination of these leaflets demonstrated connective tissue without inflammatory reaction.

Discussion

Ebstein's anomaly of the tricuspid valve is a relatively uncommon congenital heart defect showing distinct clinical manifestations with a high mortality rate in the neonatal period[2,4]. The basic defect is marked by variable degrees of displacement of proximal attachments of the valve from the atrioventricular ring[5-9]. This is associated with considerable variability in clinical outcome, with mortality rates for neonatal presentation ranging from 27% to 48%[1,10-13]. The majority of these infants die within the first week of life, although there remains a significant risk of late sudden death with or without surgical intervention[5].

The natural history of this malformation was previously based on clinical and angiographic diagnosis of older children and adults; however, echocardiography facilitated fetal and neonatal diagnosis and has redefined the outcome[14]. In severe cases of Ebstein's anomaly, cyanosis that results primarily from a right-to-left shunt at the atrial level is a prominent finding. Presence of increased pulmonary vascular resistance is an additional problem in the neonatal period. As the right ventricle cannot generate forward flow through the pulmonary arteries with increased vascular resistance, it results in functional pulmonary atresia that worsens the clinical outcome[3,5]. Mortality rate for cyanotic neonates with Ebstein's anomaly (47%) has been reported to be significantly higher than in those without cyanosis (14%)[5]. Other predictors of neonatal mortality include tethered distal attachments of the anterosuperior tricuspid leaflet; right ventricular dysplasia; left ventricular compression by right heart dilatation; atrial septal defect of more than 4 mm; and the area of combined right atrium and atrialized right ventricle greater than the combined area of the functional right ventricle.

left atrium and left ventricle[11]. Our patient had massive right atrial dilatation and a wide atrial septal defect, causing the early mortality.

Early presentation is frequently associated with other cardiac lesions, usually pulmonary stenosis or atresia. Right ventricular outflow tract obstruction is a known risk factor for early death[14,15]. In patients with severe Ebstein's anomaly and functional pulmonary atresia, the clinical differentiation between functional and structural right ventricular outflow tract obstruction needs to be clarified, but it is frequently a difficult tast[16]. Pulmonary valve regurgitation as seen on aortography or left ventriculography does not ensure a non-obstructive pulmonary valve. Rarely regurgitation may be demonstrated across a severely stenotic pulmonary valve. However, significant pulmonary regurgitation (as assessed from opacification of the right atrium on aortography/left ventriculography) with normal or dilated pulmonary root precludes the diagnosis of structural atresia or severe obstruction, as in our patient[16]. The mechanism of regurgitation of contrast material across the pulmonary valve in patients with functional pulmonary atresia is uncertain. We think the thickened nodular leaflets and annular dilatation in our patient may be the most probable causes.

The postmortem pathologic examination of our patient revealed pulmonary valve stenosis with nodular thickening of the tricuspid and pulmonary valve leaflets. Pathologic findings mentioned in other studies included significant thinning of the right ventricular free wall distal to the tricuspid valve, reduced right ventricular fiber diameter, and increased fibrous tissue content of both right and left ventricular free walls[10]. The pulmonary valve anatomy may be variable in Ebstein's anomaly with functional pulmonary atresia. Among the necropsies of Freedom et al.[16], in some of the patients the pulmonary valve appeared entirely normal, whereas in others it was redundant. In one patient the valve was congenitally bicuspid but non-obstructive. Nodular changes on pulmonary or tricuspid valves were not mentioned. Therefore this morphology observed in our patient may be another variety of the pathology.

Grant[17] reported Ebstein's anomaly and tricuspid atresia in siblings. The presence of these two distinct pathologic variants of tricuspid valve malformations in siblings caused the authors to postulate that these malformations result from a common abnormality occurring during the development of the inlet portion of the ventricle. Bharati et al.[18], in an anatomic study of cases of pulmonary atresia with intact ventricular septum, found that the tricuspid valve and the right ventricular morphology differed greatly between type 1 and type II pulmonary atresium Type II (pulmonary atresia with tricuspid insufficiency) had a greater morphologic resemblance to Ebstein's disease with pulmonary atresia than type I (pulmonary atresia without tricuspid regurgitation). The variety of morphologies within a common embryological origin versus the morphological similarities between different embryological origin suggest that many aspects of these anomalies yet to be discovered and are imprecise. In conclusion, in Ebstein's anomaly, fetal and neonatal presentation is associated with poor outcome that can be predicted with the presence of associated lesions. It probably exhibits different undiscovered anatomical morphologies, which may, in the future, enlighten the developmental pathology linked to this anomaly.

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