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Ebstein's anomaly: A Case Report

CASE REPORT

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Abstract

Introduction: Ebstein's anomaly is a rare malformation that corresponds to less than 1% of all congenital heart anomalies. It consists in the caudal displacement of the tricuspid valve with retrograde flow to the right atrium due to valvular insufficiency and it is characterized by a variable spectrum of severity, being higher in the neonatal period.

Objective: to report an Ebstein's anomaly case which early diagnosis avoided iatrogenic ducts.

Case report: a newborn at term, appropriate weight for gestational age, female, was born of natural childbirth, with Apgar score 8/9 from pregnancy without complications. At birth, not in need of resuscitation in the delivery room, but presented heart murmur and fall of saturation, being supported and then forwarded to the NICU. Not present hemodynamic instability. The chest x-ray showed increased cardiac area with increased right atrium. The Transthoracic Echocardiogram showed mild right ventricular dilatation and important of the right atrium, tricuspid valve dysplasia with low implantation of posterior leaflet of tricuspid insufficiency presence important to Doppler, being diagnosed with Ebstein's anomaly. Evolved with progressive improvement of the frame and saturation above 95% on room air to pulse oximetry. Patient follows in outpatient follow-up in use of inotropic and diuretic, remaining asymptomatic.

Conclusion: the recognition of the gravity of the picture is relevant to the proper management in order to prevent iatrogenic ducts, which can lead to complications or permanent sequelae.

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Ebstein's anomaly; congenital heart disease; Tricuspid insufficiency; iatrogenesis.

Introduction

Ebstein's anomaly is a rare heart disease that variable morphology corresponds to less than 1% of all congenital cardiac anomalies. In the general population, the incidence corresponds to 1/210,000 live births, with the male-to-female ratio of 1:1. There are cases in which there is relation with family history, however the most is sporadic[1,2].

It is characterized by malformations and low deployment of septal and posterior leaflets of the tricuspid valve, showing an area of "atrialized" right ventricle[3]. The etiology of this anomaly can be explained by the rupture of the papillary muscle of the tricuspid valve. The typical presentation of Ebstein's anomaly in the neonatal period is cyanotic newborn with cardiomegaly to x-ray[4].

In the initial investigation of these patients, cardiac acute clinical examination, chest x-ray and eletrocardiography can be useful[5]. However, the echocardiography, in most cases, is the best test for confirmation of the diagnosis by allowing the quantification of severity and prognosis of malformation[6,7].

The occurrence of severe tricuspid insufficiency in newborns is poorly tolerated, since the pulmonary vascular resistance is high at birth, being the most serious appearance of this anomaly in the neonatal period. Mortality is approximately 50% of cases in the sequence of natural history, and about 30% of patients in this age group[8,9].

The newborn that exhibits cyanosis is well handled by conservative techniques until the pulmonary pressure decline. Each patient's therapy is adapted according to the severity and degree of functional obstruction of the outlet of the right ventricle. The infusion of prostaglandins and nitric oxide can be advantageous in more severe patients[1]. One of the interventions for the surgical correction of that heart disease is the "cone technique", developed since 1989 by da Silva et al.[9,10].

The aim of this study is to report an Ebstein's anomaly case which early diagnosis avoided iatrogenic ducts.

Case report

Newborn at term, appropriate for gestational age, female, was born of natural childbirth, with Apgar of 8/9 from pregnancy without complications. At birth, not in need of resuscitation in the delivery room, but presented heart murmur and fall of saturation, being offered supplemental oxygen in Hood to 40% and then forwarded to the NICU. In the NICU clinical evolution, the newborn not present hemodynamic instability. The chest x-ray showed increased cardiac area with increased right atrium. Transthoracic echocardiogram showed patent ductus arteriosus and patent foramen ovale, *situs solitus*, atrioventricular and arterial ventricle consistent connection, moderate right ventricular dilatation and important of the right atrium, dysplastic tricuspid valve with low implantation of posterior leaflet of tricuspid insufficiency presence important to Doppler, being diagnosed with Ebstein's anomaly. Evolved with progressive improvement of the frame and saturation above 95% on room air to pulse oximetry. On discharge, was prescribed propranolol and furosemide on function of heart failure present. Patient follows in outpatient follow-up in use of these drugs remain asymptomatic.

Discussion

Ebstein's disease is a cardiac anomaly with variable clinical conditions that is characterized by apical displacement of the septal and posterior leaflets of the tricuspid valve. The morphology of this condition leads to decrease the size of the functional ventricle. The region above the tricuspid ring displaced

is described as an "atrialized" region of the right ventricle. It has thinner wall assuming similar aspect atrial anatomy, which is partly responsible for defect of ventricular filling[1,2].

The natural history of the disease depends on the severity, considering a variety of cases. In more severe cases, cyanosis and heart failure happens in the first month of life[1]. As the pulmonary vascular resistance decreases, due to change in fetal circulation pattern, there is a tendency of improvement of clinical conditions[11]. In milder cases of the disease, diagnosis is difficult and can go undetected for many years because patients initially may be asymptomatic. However, some phenomena can be purchased along the natural history of this anomaly, such as volume overload and right ventricular dysfunction[12].

Some abnormalities may be associated with Ebstein's anomaly, including: atrial septal defect in 90% of patients, pulmonary stenosis and pulmonary atresia. In small percentage of cases and occasionally can also occur ventricular septal defect[1,13,14].

It is cyanotic anomaly where the newborn is well managed by conservative techniques to pulmonary pressure decline. In severe cases, a constant intravenous infusion of prostaglandin E enables the maintenance of patency of the ductus arteriosus and ensures adequate pulmonary blood flow[15,16].

For this reason, facing any cyanotic newborn with heart disease and unfavorable outcomes, initial management includes ensuring central venous access route, infusion of prostaglandins and ventilatory support[4]. Inhaled nitric oxide administration may also be useful due to its role as a selective pulmonary vasodilator in persistent pulmonary hypertension of the newborn[17].

In the reported case, the patient had a heart murmur and fall saturation after birth. Therefore, oxygen support was carried out with 40% Hood, noting improvement, without further measures. Oxygen therapy, carried out in the case, aimed to promote the reduction of pulmonary vascular resistance by increasing arterial oxygen saturation and, consequently, due to its vasodilating action. These factors are essential in improving gas exchange and the disappearance of cyanosis[11]. Thus, it is worth emphasizing the importance of recognizing the improvement of cyanogenic frame, from simple measures, thus avoiding iatrogenic behavior.

The patient with congenital heart disease may present important gravity in the neonatal period. This gravity will be influenced by physiological changes typical of adaptation to extra-uterine environment and the importance of cardiac defects present. The clinical cardiologic examination, chest X-ray and electrocardiogram may be useful in the initial investigation of these patients. However, the echocardiogram often is the best test to confirm the diagnosis[6,7].

Performing Doppler / Echocardiography is relevant to anatomical assessment of the presence of obstructive lesions of the right heart. However, due to an elevated pulmonary vascular resistance, there may be difficult to differentiate functional or anatomical obstruction[15].

In the investigation of the present case, the chest X-ray revealed an enlarged heart with enlarged right atrium **(Figure 1)**. Which, together with the initially unfavorable evolution of the clinical picture leads to suspicion of Ebstein's anomaly. The diagnosis was confirmed in the first hours of life, with the completion of transthoracic echocardiography to bedside. This was evidenced characteristic image of dysplasia and caudal displacement of the tricuspid valve, and presence of moderate dilatation of the right ventricle, significant dilation of the right atrium and the presence of tricuspid insufficiency to doppler **(Figure 2)**.

The surgical interventions in Ebstein's anomaly are considered on the following indications: symptomatic patients in functional class III and IV of the New York Heart Association (NYHA); in functional clas-

Figure 1: Chest X-ray incidence anteroposterior demonstrating increased cardiac area.

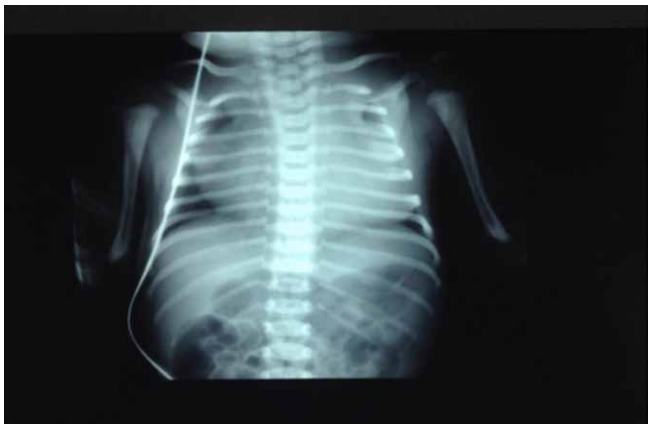
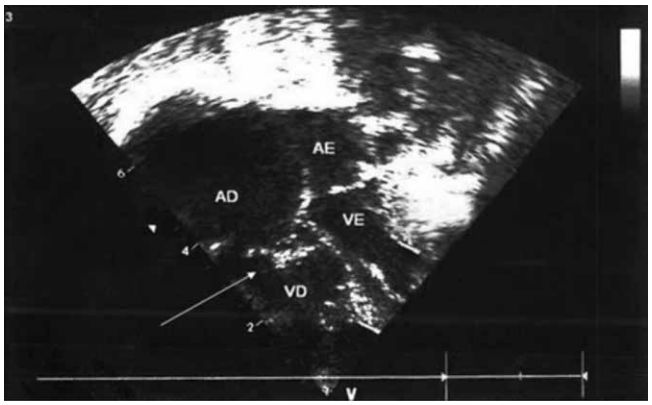


Figure 2: Bidimensional echocardiography, four-chamber plane. Cut demonstrating caudal displacement of the tricuspid valve (white arrow), with low implantation of posterior leaflet, moderate dilatation of the right ventricle and significant dilation of the right atrium.



ses I and II, but with cardiomegaly, a cardiothoracic index of 0.65 or greater; significant cyanosis and polycythemia; events of paradoxical embolism; uncontrollable tachycardia and atrioventricular bundle accessory[18].

The surgical procedure should be carried out taking into account the anatomy and the probability that it will be successful, since studies showed up to 47% mortality level in operated neonates. The repair of the valve in a gravely ill newborn with low right ventricular function is generally unwise, given the variability of anatomy and unpredictable results[15, 19].

To prevent the occurrence of right heart failure is required that the atrial septal defect is left open in order to allow the right to left shunt. When the repair is done, there are variety of surgical options for treating valvular malformation[15].

Among the surgical procedures, the Blalock-Taussig surgical modified associated with ligature of the main pulmonary artery technique can be initially considered the best palliative option for children who have pulmonary atresia with severe tricuspid insufficiency and pulmonary insufficiency[19]. According Shinkawa et al[20], in a study with 40 patients with symptomatic anomaly of Ebstein in neonatal period[16], patients did not undergo surgery at this time. These show survival rate of 94 %. While in 9 patients operated in the neonatal period using the Blalock - Taussig modified technique, the survival rate at ten years old was 76.2 %[9]. This study concluded that there is a benefit in early infusion of prostaglandins without performing surgery. In patients unable to be weaned prostaglandin a systemic-pulmonary shunt is an alternative[19].

Among other procedures to correct this disease, there is the "cone" technique. This technique proved be the lowest mortality and present best results. However, it has relative contraindication to use in newborns[9]. The technique consists in positioning the valves of the tricuspid valve in the true ring and longitudinal plication of the "atrialized" right ventricle thus maintaining the size and appropriate morphology of the right ventricle[10].

This report demonstrates the importance of recognizing the severity of the anomaly, essential for carrying out appropriate measures for each case. Up to 50% cases can be fatal, especially when symptomatic in neonatal period. There is a tenden-

cy toward invasive measurements in these patients, which may be unnecessary, causing various complications or permanent sequelae[16,21,22].

Although often be necessary to use invasive measures such as, for example, conventional mechanical ventilation. There is increasing evidence that lung function may worsen and contribute to the development of multiple organ dysfunction. Among some complications, continuous positive airway pressure is related to decrease cerebral perfusion due to increased average pressure and intracranial pressure. Others that can be cited are: trauma airway, nosocomial infections and bronchopulmonary dysplasia[23-27].

Conclusion

Early diagnosis of Ebstein's anomaly is of fundamental importance for the prognosis of the patient, because 50% of cases can be fatal, especially when symptoms occur during the neonatal period.

In this patient was not necessary to adopt invasive measures or reconstructive surgery. The recognition of the gravity of the picture is relevant to the proper management[28] in order to prevent iatrogenic ducts, which can lead to complications or permanent sequelae.

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