Ebstein's anomaly revealed by hydrops fetalis : a case report

Bellalah. M [1,4], Methlouthi. J [1,4], Bouabdallah. O [1], Mghirbi. O [1,4], Braham. D [1], Barka. M [1], Ghaith. A [1], Bouhlel. I [2,4], Masmoudi. A [3], Ben Jamaa. N [3], Nouri. S [1,4], Mahdhaoui. N [1,4]

[1] Department of Neonatology, Farhat Hached Hospital,Sousse ,Tunisia
[2] Department of Cardiology, Farhat Hached hospital, Sousse,Tunisia
[3] Department of fetopathology, maternity and neonatology center of Tunis, Tunisia
[4] Faculty of Medicine of Sousse, University of Sousse, Laboratory LR14ES05

ABSTRACT

Ebstein's anomaly is a congenital heart defect rarely revealed by foetal anasarca. We report an original case of Ebstein's disease revealed by hydrops fetalis in a preterm newborn. The antenatal ultrasound revealed anasarca, ascites, pericardial effusion, cardiomegaly and detachment of the scalp. Echocardiography and clinical findings led to a preliminary diagnosis of non-immune hydrops associated with congenital heart disease.
The diagnosis of Ebstein anomaly was made and confirmed by post mortem anatomopathological examination.

Keywords : hydrops fetalis, Ebstein, neonatal

INTRODUCTION

Ebstein's anomaly (EA) is a rare complex congenital heart disease (CHD) of the tricuspid valve, first described by Wilhelm Ebstein in 1866. It concerns less than 1% of CHD (1). Fetal diagnosis and neonatal presentations of the disease are the most severe and associated with the highest mortality rates (2). Cardiac etiologies are responsible for 10% to 20% of cases of non-immune hydrops fetalis (NIHF).We report an original case of EA revealed by hydrops fetalis in a preterm newborn.

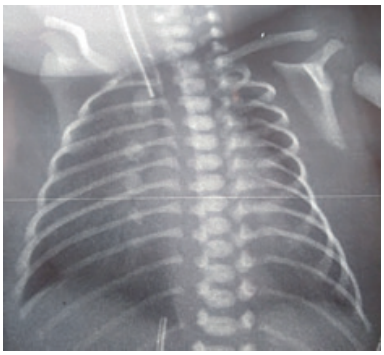
CASE REPORT

A premature male newborn was the first child of young consanguineous parents.
The mother was 18 years old. There was no infectious history. Pregnancy was complicated with a diagnosis of hydrops fetalis at 26 weeks of gestational age (GA). Antenatal ultrasound performed at this term revealed: hydramnios, ascites, pericardial effusion, cardiomegaly and detachment of the scalp. Maternal TORSCH serologiy screen was negative. Doppler ultrasound did not show signs of anemia. The delivery was performed vaginally at 30 Weeks GA. Apgar score was 0 and 7 at 1 and 5 minutes respectively. Initial and active resuscitation was performed.
Abdominal paracentesis was performed draining 150 ml of a transparent and tinged yellow fluid.
Physical examination on admission found a eutrophic child with birth weight of 2045g (50th percentile), facial dysmorphia, generalized skin edema, a systolic murmur, weak femoral pulse, distended abdomen with moderate hepatomegaly and edema of external genitalia. Blood pressure was 40/20 mmHg. His ventilator needs were increasing and percutaneous oxygen saturation was 50%. Blood gas showed severe respiratory acidosis. His blood group was A (Rh+) with negative coombs test. Aspartate transaminase (AST) was72 u/l and alanine transaminase (ALT) was 7 u/L. The level of total protidemia was of 60 g/l. The ascitic

Corresponding author :
Dr. Bellalah Manel
Address : Department of neonatology.
Farhat hached hospital. Rue Ibn Jazzar. 4031 .SOUSSE .EZZOUHOUR SOUSSE. Tunisie.
Phone : 00 216 21 701 369
Mail : bellalahmanel@yahoo.fr

fluid contained 150 leukocytes/µL with 100% lymphocytes, 550 red blood cells/µl, and a protein level of 10g/l. He was started on intravenous antibiotics because of the severity of the features.

Chest X ray showed cardiomegaly with cardiothoracic ratio at 0.78 with apparently normal pulmonary parenchyma and no signs of respiratory distress syndrome (RDS)) (Figure1).



Figures 1 : Chest X Ray showed an important cardiomegaly (ICT =0.78)

Initial echocardiography revealed left ventricle dysfunction with ejection fraction of 50%, a huge right atrium, four chamber views of the heart showed marked downward displacement of posterior leaflet of the tricuspid valve with attachment to underlying free wall, markedly dilated atrialized portion of right ventricle (ARV), small functional portion of right ventricle (RV), and dilatation of right atrium (RA). Tricuspid insufficiency, pulmonary hypertension at 50mmhg, a small patent foramen ovale, moderate pericardial effusion and persistent ductus arteriosus (PDA) and no aortic coarctation. Severe Ebstein anomaly was suspected (Figure 2).

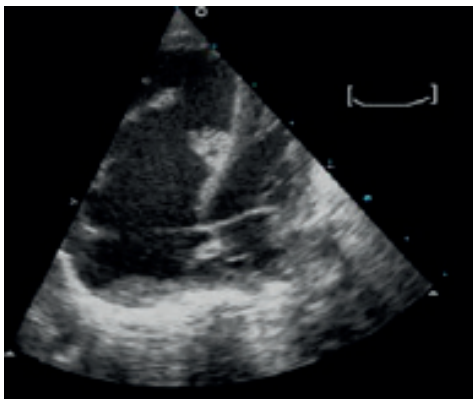
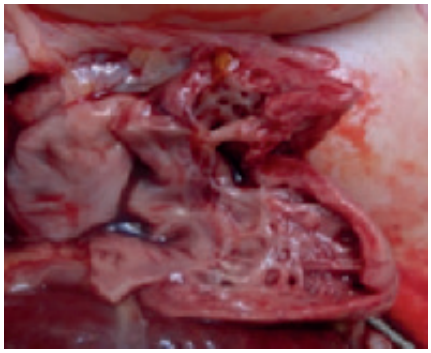


Figure 2 : Echocardiography : four chamber views of the heart with severe Ebstein's anomaly showing marked downward displacement of posterior leaflet with attachment to underlying free wall, markedly dilated atrialized portion of right ventricle (ARV), small functional portion of right ventricle (RV), and dilatation of right atrium (RA).

The first diagnostic hypothesis was of a cardiogenic shock. Fluid restriction was instituted. A management with amines (Dobutamin (10ug/kg/mn)) was added. The anemia was corrected and the neonate received platelet transfusion and fresh frozen plasma (FFP) because of umbilical hemorrhage. About five hours after admission, he presented an acute cardio-circulatory failure and Clinical and ultrasound findings had led to a preliminary diagnosis of NHIF associated with CHD. Autopsy was indicated and was performed after a written consent from parents. The post mortem anatomopathological examination concluded to Ebstein's disease. The posterior and septal tricuspid leaflets were displaced downward and were dysplastic. The atrialized right ventricular chamber was dilated. There was also an enlarged right atrium and a patent ductus arteriosus (Figure 3).



Figures 3 : Four chamber views and autopsy of the heart with severe Ebstein's anomaly showing marked downward displacement of posterior leaflet with attachment to underlying free wall, markedly dilated atrialized portion of right ventricle (ARV), small functional portion of right ventricle (RV), and dilatation of right atrium (RA).

The final diagnosis was of NHIF caused by a severe Ebstein's anomaly.

DISCUSSION

Hydrops fetalis is associated with a pathologic increase in interstitial and total fetal body water, usually appearing in foetal soft tissues and serous cavities (3). Its definition is based on ultrasound findings: it is the combination of a subcutaneous edema, greater than 5 millimeters and an effusion in a serosa (pericardium, pleura or peritoneum) or the presence of serous effusion in two sera at least without subcutaneous edema. Since widespread prevention of hydrops fetalis caused by Rhesus immunization, over 90% of cases of hydrops are not immune (3-4) and cardiovascular abnormalities became the most common underlying cause of hydrops (5).

Cardiac etiologies are responsible for 10% to 20% of cases of non-immune hydrops fetalis (NIHF).The cardiac structural malformations are the most associated with hydrops (5).

Ebstein's anomaly (EA) is rarely revealed by hydrops (6). EA is a special form of tricuspid valve (TV) dysplasia, characterized by the downward displacement of

septal leaflet and atrialized right ventricle (RV) (7). EA is a special form of tricuspid valve (TV) dysplasia (8, 9);there is incomplete delamination of the posterior and septal leaflets of the TV. The normal process of delamination continues into the second trimester, although it is complete for the anterior leaflet much earlier, usually during the first trimester (10, 11).
The clinical characteristics of EA are twofold: redundancy of some of the TV leaflets, and varying degree of adherence of TV leaflets to the free wall of the right ventricle (RV). This results in "apical displacement" of the TV into the RV to a variable degree, with valvular incompetence, a myopathic RV, and right heart failure. Most cases of EA are sporadic; familial cases are rare. There is a higher incidence of EA occurring in babies of mothers who received lithium during pregnancy. In our case; a history of medication taken during pregnancy was not noticed.

EA has an extremely variable natural history. It may present at any age, with a variety of hemodynamic and electrophysiologic sequelae. If the deformity of the tricuspid valve is severe, neonatal congestive heart failure, arrhythmia leading to foetal hydrops or even intrauterine death may occur. In contrast, patients with isolated minor tricuspid valve displacement may remain asymptomatic until late adult life.
The hemodynamic variations and clinical presentation depend on age at presentation; anatomic severity, hemodynamics, and degree of right-to-left inter atrial shunting (9).

Ebstein's anomaly usually presents with hemodynamic sequelae of severe tricuspid valve displacement and right ventricular abnormalities and has a poor prognosis. There is an early death hazard due to heart failure and intrauterine cardiomegaly causing pulmonary hypoplasia. Hemodynamic deterioration may be due to increased right to left shunting and right or left heart failure, or both. Although, left ventricular dysfunction is especially difficult to assess in EA because of paradoxic septal motion, which influences the estimation of ejection fraction and fractional shortening. Symptoms may be exacerbated by prematurity and their high pulmonary vascular resistance, like in our observation (10).
Echocardiography is the most important diagnostic test in Ebstein's anomaly, permitting accurate assessment of the anatomy and distal attachments of the tricuspid valve, the size and contractility of the functional right ventricle and the overall grade of disease severity.

EA in our patient was highly suspected during the etiological approach of hydrops fetalis. The diagnosis was confirmed by post mortem anatomopathological examination. The lack of performance of our bedside ultrasound machine did not help us make the diagnosis before death.

The severity of the disease is described in the classification of Carpentier. It indicates the following types: -Type A: minimal displacement of septal leaflet attachment with small atrialized right ventricle (RV)
-Type B: moderate displacement of septal leaflet attachment with large atrialized RV
-Type C: important displacement of septal and postero-inferior leaflet attachment with non atrialized or dyskinetic atrialized RV, restrained anterior leaflet motion, and short chords; and -Type D: tricuspid sack (9,11).
Our patient had a probable Ebstein's disease type C with significant tricuspid regurgitation, a patent ductus arteriosus and small patent foramen ovale.
The support varies depending on the severity of cardiac involvement. Ebstein's anomaly is a ductal-dependent heart disease. The management of this disease is based on the treatment of right heart failure. Treatment may involve Prostaglandin E1 (PGE1) which can increase pulmonary artery pressure (12).Patients with Ebstein's anomaly and cardiac failure who are not candidates for surgery, like our patient are treated with standard heart failure therapy, including diuretics and cardiotonics.

Multiple different techniques, surgical and interventional catheterization based, have been used to obtain adequate pulmonary blood flow and limit cardiac failure. Surgical operation is indicated for patients with Ebstein's anomaly in the presence of right heart dilation and progressive ventricular functional impairment (13). However, the optimal management strategy for symptomatic neonates with EA remains unknown.

Predictors of a bad outcome are severe tricuspid regurgitation, significant cyanosis (oxygen saturation <85%), cardiomegaly (cardiothoracic ratio>0.8 by chest radiograph) a large PDA and small patent FO (14, 15).Our patient presented with all those poor factors.

Patients with Ebstein's anomaly also have conduction system abnormalities, which are at least partly due to the compression of the AV node by the septal malformation, accessory pathways, and abnormalities of the right bundle branch.
Mortality has previously been reported to be as high as 47% to 70% in symptomatic neonates who are admitted with diagnosis of EA, and close to 100% in those subsets of neonates with severe cyanosis
In our case the prognosis was interdisciplinary estimated as being very poor.

CONCLUSION

Prognosis of NIHF differs between different etiological groups. It is essential to identify the etiology to better prognosis, provide appropriate treatment, and assess recurrence risk for future pregnancies. In EA, fetal and neonatal presentation is associated with a poor outcome and can be predicted by the echocardiographic appearance and presence of associated lesions. Therefore, autopsy should be recommended in all cases of unexplained NIHF and in cases of fetal or neonatal death or pregnancy termination in order to determine a probable recurrent etiology in future pregnancies..

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Authors' contributions : All the authors had contributed to acquisition of data,to the management of the patient; drafted the article or revising it ; and gave final approval of the version to be published.

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