Prenatal diagnosis of ebstein anomaly – from normal to severe disease

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Objective

Ebstein anomaly is a congenital malformation characterized primarily by abnormalities of the tricuspid valve and right ventricle (Figure 1), with an incidence of 1/20000 live births, widely variable clinical presentation and spectrum, and in extreme cases it constitutes a ductus dependent heart disease, hence the importance of prenatal diagnosis.

Methods

Presentation of one case and literature review.

Results

A 26-year-old woman, gravida 2, parity 1, at 31 weeks gestational age, without any fetal abnormalities detected during her pregnancy, including her first and second trimester routine ultrasound scans, was diagnosed with fetal cardiomegaly on her third trimester routine ultrasound scan. Fetal echocardiogram revealed an Ebstein anomaly (EA) of the tricuspid valve, with severe tricuspid regurgitation, right atrium (RA) dilation, severe cardiomegaly and no anterograde flow through the pulmonary valve, with reverse flow in the ductus arteriosus. A forceps delivery was performed at 38 weeks and 2 days of gestation. A female neonate was born weighting 2625 grams, with an APGAR of 7/8/9. She had an episode of respiratory distress with bradycardia at 2 minutes of life with need of positive pressure ventilation and intubation. The baby was transferred to the Neonatology Intensive Care Unit where she was started on Prostaglandin E2 infusion. A chest radiography showed severe cardiomegaly and bilateral pleural effusions. A postnatal echocardiogram confirmed the EA with apical displacement of the septal leaflet of the tricuspid valve, without coaptation of the leaflets, conditioning severe regurgitation and RA enlargement and cardiomegaly. She had no anterograde flow from the right ventricle to the pulmonary artery through the pulmonary valve. The pulmonary territory was being irrigated by the left-right shunt through the large arterial duct and an atrial septal defect with right-left shunt. A left thoracocentesis was performed and diuretics were initiated resulting in total resolution of pleural effusions and improvement of cardiomegaly. She was transferred to a heart surgery centre and proposed for palliative surgery - Blalock Taussig Shunt - but serial echocardiograms showed, with the progressive weaning of prostaglandins, a gradual improvement of the right ventricular function, with anterograde flow through the pulmonary valve, ensuring an acceptable oxygen saturation on the 19th day of life when she was discharged without surgical correction. At 2 months of life she has a good clinical situation.

Conclusion

Ebstein anomaly is characterized by variable malformed and displaced tricuspid valve leaflets that are partly attached to the tricuspid valve annulus and partly attached to the right ventricular endocardium. These features cause tricuspid valve regurgitation and right heart enlargement and, in the most severe cases, the absence of anterograde flow through the pulmonary valve may exist and lead to ductus dependent heart disease requiring intensive care and heart surgery in the neonatal period. In this case the baby fortunately had a prenatal diagnosis: although she needed mechanical ventilation and prostaglandins, she recovered the ventricular function and the flow through the pulmonary valve and did not require surgery. Echocardiography is generally the key test for the diagnosis and initial anatomic evaluation of Ebstein anomaly. As observed in our case, the clinical features may be absent until late in pregnancy. It is important to detect, suspect and investigate further whenever the first signs appear on a routine ultrasound scan as the mortality decreases significantly with appropriate prenatal and neonatal management.
Apical displacement (*) of the septal leaflet of the tricuspid valve and the displaced, long (sail-like) anterior leaflet.