Rare disease

Fetal Ebstein's anomaly

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DESCRIPTION

A 36-year-old Gravida 2, Para 1 woman at 34 weeks' menstrual age was referred for a fetal echocardiogram in view of fetal hydrops detected by obstetric sonography. She gave a history of consanguineous marriage and her first child was healthy.

Fetal echocardiogram (four-chamber view) performed revealed findings consistent with Ebstein's anomaly; a downward displacement of septal leaflet of the tricuspid valve, which were tethered to the muscular part of ventricular septum, right ventricular atrialisation, a small effective right ventricular cavity and cardiomegaly (figure 1). The echocardiogram also showed severe tricuspid valve regurgitation (TR).

A full-term male neonate was born cyanotic with an Apgar score of 7 and SaO₂ of 82% on room air. An echocardiogram obtained on the first day of life, showed Ebstein's anomaly with severe tricuspid regurgitation (figure 2). There was no associated right ventricular outflow tract obstruction or pulmonary atresia (figure 3).

Patent foramen ovale with right to left shunting was also noted. Apical displacement index (normal 8 mm/m²) was 27 mm/m². Celermajor index (Gose score) (ratio of the combined area of the right atrium and atrialised right ventricle to that of the functional right ventricle and left heart in a four-chamber view at the end-diastole) was 0.98 (grade 2). This ratio has four grades: ratio <0.5=grade 1; ratio 0.5–0.99=grade 2; ratio 1–1.49=grade 3 and ratio >1.5=grade 4. It is a prognostic tool: when the ratio is >1.5, 100% mortality is likely and a ratio <1 indicates 92% survival. Right atrium diameter at interatrial septum was 30 mm and antero-posterior was 23 mm. Biventricular ejection fraction was normal (left ventricular ejection fraction=66%, right ventricular ejection fraction=64%). Tricuspid regurgitation pressure gradient was 43 mm Hg.

Ebstein's anomaly is a rare congenital cardiac malformation (prevalence 0.3–0.7% of all patients with congenital heart disease¹); characterised by varying degrees of downward displacement of the tricuspid valve leaflets into the

Figure 1 Fetal echocardiogram (four-chamber view) (left) shows downward displacement of septal leaflet of the tricuspid valve, right ventricular atrialisation, a small effective right ventricular cavity and cardiomegaly. Blue oblique line denotes the length of STL displacement from AML. Image on right shows severe tricuspid valve regurgitation on colour flow doppler. AML, anterior mitral leaflet; LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle; STL, septal tricuspid leaflet and TR, tricuspid regurgitation.

Figure 2 Neonatal echocardiogram (four-chamber view) (left) shows downward displacement of septal leaflet of the tricuspid valve, right ventricular atrialisation, a small effective right ventricular cavity and cardiomegaly. White oblique line denotes the length of STL displacement from AML. Blue arrow indicates a patent foramen ovale. AML, anterior mitral leaflet; STL, septal tricuspid leaflet.

cavity of the right ventricle. Fetal and neonatal presentation in Ebstein's anomaly is predicted to have a poor overall prognosis^{2 3} as the age at presentation is dependent on the severity of anatomic and haemodynamic derangements. In utero presentation with fetal hydrops and neonates with cardiomegaly, congestive heart failure and cyanosis have a poor outcome. Echocardiographic predic-

tors of severity include the degree of tricuspid valve deformity, insufficiency and displacement, right-sided chamber dilatation, associated right ventricular outflow tract obstruction and a significant right-to-left shunting. Differential diagnosis for fetus and neonates with dilated right atrium and TR include pulmonary atresia, isolated congenital tricuspid insufficiency due to tricuspid valve dysplasia or prolapse and Uhl's anomaly. Targeted fetal echocardiography is sensitive and specific for antenatal diagnosis of congenital heart disease in high-risk pregnancies.

Figure 3 Neonatal echocardiogram (short axis view) shows the displaced septal tricuspid leaflet towards the RVOT with normal pulmonary artery and leaflets. Ao, aorta; PA, pulmonary artery; RV, right ventricle; RVOT, right ventricular outflow tract and STL, septal tricuspid leaflet.

Learning points

- ▶ Ebstein's anomaly, a rare congenital cardiac malformation, is associated with a worse prognosis when it presents early as heart failure in the fetus and neonate.
- ▶ Targeted fetal echocardiography helps in risk stratification of such high-risk pregnancies towards more aggressive management.

Competing interests None.

Patient consent Obtained.

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