17th World Congress in Fetal Medicine

A case of pentalogy of cantrell with cystic hygroma diagnosed in the first trimester

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Objective

Pentalogy of Cantrell is a rare congenital syndrome first described by Cantrell in 1958. Thoracoabdominal variety of ectopia cordis accounts for only 7% of cardiac ectopies, which is known as pentalogy of Cantrell. At prenatal ultrasound the entity should be considered if the heart is seen outside the cavity of the chest (ectopia cordis) and within the exomphalos. Fetuses affected by pentalogy of Cantrell may manifest fluid accumulation in the neck or chest. Here we present a case detected in first trimester screening ultrasound.

Methods

This is a case report.

Results

A41-year-old pregnant woman G2P1 underwent a first-trimester screening for aneuploidy. The scan demonstrated an alive fetus of 66 mm crown-rump length consistent with 13 gestational weeks. An exomphalos measuring 19x16mm was protruding from the anterior abdominal wall. Evisceration of the heart, liver and major parts of intestinal loops were seen. Due to early gestational age, no intracardiac abnormality was detected. Color Doppler sonography clearly showed the fetal heart pulsating outside of the thorax. The nuchal translucency thickness was significantly increased (11. 6mm) associated with thin internal septations. The parents opted for elective termination of the pregnancy and after delivery the pentalogy of Cantrell was confirmed in the male fetus.

Conclusion

Diagnosis of pentalogy of Cantrell using 2 dimensional and Doppler sonography is possible in the routine first trimester examination. Cystic hygroma is not only associated with chromosomal abnormalities but also associated with other fetal malformations, mostly cardiac anomalies such as this syndrome. To differentiate pentalogy of Cantrell from other abdominal wall defects, it is important to pay attention to umbilical cord insertion, organ contents of the exomphalos, associated anomalies and presence or absence of bands and membranes.