18th World Congress in Fetal Medicine

The variable ultrasound features in the semilobar form of holoprocencephaly

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Objective

Holoprosencephaly has evident features in its alobar form. Semilobar and lobar forms present with milder ultrasound characteristics. In this case we present a case of the semilobar form depicting its ultrasound characteristics.

Methods

We report a case of a 28 years old G2 P1 with no particular history. First trimester NT was normal but showed abnormal features at the central nervous system. She was referred for additional ultrasound evaluation at 14 weeks of gestational age.

Results

A unique ventricular cavity joined both hemispheres. A single thalamus was identified at the midline with a partial division seen in almost the third of it and it was not possible to identify a 3rd ventricle. Two choroid plexuses were identified, one in each side of the ventricular cavity. Mild hypotelorism was observed along with a flat aspect of the face, without a proboscis. There were no other abnormalities identified, mainly no omphalocele, diaphragmatic hernia, limb, or kidney abnormality. Heart was in normal position with a cardiac axis of 47 degrees and a normal four chamber view. Biometry was at the 50th centile and the movement of the fetus was within normal. All the features were discussed with the parents along with the possible diagnosis. The couple decided for a medical termination of the pregnancy.

Conclusion

Holoprosencephaly carries a poor prognosis. Milder forms present with milder ultrasound features, yet the parents should be informed about the diagnosis as early as the first trimester to allow them to take appropriate decisions in this early gestational age.