A case of holoprosencephaly

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Objective

Holoprosencephaly (HPE) is a developmental abnormality characterized by congenital forebrain and midline-face malformations. The prevalence of HPE is estimated to be 1 per 16,000 live births. An abnormality of chromosome number is overall the most frequently identified etiology in a patient with HPE. These abnormalities include trisomy 13, trisomy 18, and triploidy, though several others have been reported. Based on severity, HPE is classified into four types such as HPE, semilobar HPE, lobar HPE, and middle interhemi-spheric variant (MIHV). In addition, holoprosencephaly associated with hydrops fetalis have been very few cases reported. We report a rare case of holoprosencephaly associated with hydrops fetalis diagnosed prenatally.

Methods

Here we present the case of a 22-year-old woman, who came into the emergency department at her 20 weeks of gestation with abdominal pain. Ultrasonography showed HPE lobar and fetal hydrops.

Results

We performed an ultrasound scan with advanced neurosonography study, the cavum of the septum pellucidum is not visualized, the thalami are separated and dilatation of the third ventricle as well as hydrops fetalis, pericardial and pleural effusion were noted by level II ultrasound. Under the impression of holoprosencephaly lobar with hydrops fetalis, pericardial and pleural effusion termination of pregnancy with misoprostol was undertaken. The histopathology of fetal autopsy confirmed our diagnosis.

Conclusion

Fetus with HPE might have other associated structural abnormalities. Hydrops fetalis are rare associations. Meticulous sonographic examination to depict the associated defects are necessary in any fetus with holoprosencephaly.