A case of lobar holoprosencephaly with mega cisterna magna

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Objective

Holoprosencephaly (HPE) is the most common malformation of the prosencephalon. Its incidence is estimated to be 1 in 16000 live births and 1 in 250 spontaneous miscarriages. It can be structurally categorized in 3 types, according to the degree of cerebral involvement: alobar, semilobar and lobar. Lobar holoprosencephaly (HPE) is the least severe form of HPE and can be seen with an interhemispheric fissure present along the entire midline with differentiation of cerebral hemispheres. Here we report a case of lobar holoprosencephaly.

Methods

This is a case report.

Results

A 25 year-old patient G2P1 was referred to our perinatology clinic with ventriculomegaly. They had history of first-degree consanguinity; there was no maternal medication use or exposure to radiation during pregnancy. She did not have first or second trimester screening test. In our ultrasound examination, she had 32 week 2 day gestation, the cavum septi pellucidum was absent and the lateral ventricles appeared significantly dysmorphic and fused from the level of the frontal horns to the bodies. Ventriculomegaly and agenesis of corpus callosum were also detected. Cisterna magna was measured 17mm, the cerebellum and vermis was of normal size and structure. The family was informed of the diagnosis of lobar holoprosencephaly and its poor prognosis. Elective cesarean section was performed at 39 weeks due to transverse lie. A 3200 g male infant was delivered in good condition. No additional abnormalities were detected upon postnatal examination. In the first day of life the neonate had seizures. Transfontanel ultrasonography reported lateral ventriculomegaly and agenesis of corpus callosum. Cranial CT was planned but baby died on the postnatal third day due to rapidly progressing potassium excess, seizures and cardiac arrest.

Conclusion

Prognosis is generally poor and dependent upon the degree of fusion and malformation of the brain. Other health complications may be present. Alobar and semilobar HPE are lethal. Babies affected with HPE can survive for years, but severe neurologic manifestations and mental retardation are to be expected.