Antenatal Sonographic Detection of Holoprosencephaly

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MJAFI 2007; 63 : 88-89

Key Words : Holoprosencephaly

Introduction

Holoprosencephaly [1,2] is characterized by absence of foetal cerebral tissue derived from prosencephalon due to failure of division or incomplete cleavage and morphogenesis of forebrain. Its incidence is 1:13000. It is of three types; alobar, semi-lobar and lobar depending upon the severity of the condition. Ultrasonography (USG) is the ideal modality for antenatal detection of this condition [3]. One such case detected by USG is reported .

Case Report

A 27 year old lady reported for routine antenatal ultrasound in the fifth month of pregnancy. She had a five year old daughter from previous normal delivery. There was no contributory history of any previous congenital anomaly or any spontaneous abortions. Ultrasound revealed 17 weeks gestation of the foetus. The foetal head scan revealed an anechoic cystic cavity suggestive of a monoventricle in the cerebral region. No normal cerebral tissue was visualized, the thalami were fused together with hypotelorism. No other anomalies were noted in the foetus (Fig.1) . A diagnosis of alobar holoprosencephaly was made .

Discussion

The neural tube is formed at the end of fourth week and is divisible into an enlarged cranial part (brain) and a caudal tubular part (spinal cord). In the fifth week, the cavity of brain shows three dilatations or ventricles namely prosencephalon, mesencephalon and rhombencephalon. The prosencephalon further subdivides into telencephalon and diencephalon which together form cerebral hemispheres. The cavity of each telencephalic vesicle becomes lateral ventricle while that of diencephalon becomes the third ventricle. Complete or partial failure in division of cerebrum (prosencephalon) into hemispheres and lobes results in holoprosencephaly. There is failure of lateral cleavage into distinct cerebral hemispheres and failure of transverse cleavage into diencephalon and telencephalon.

Fig. 1 : Ultrasonography scan

Holoprosencephaly [4], usually occurs during fourth to sixth week of gestation by ventral induction during which time the prosencephalon usually divides. The aetiology remains obscure. The common associated conditions are Trisomy 13, 18 and 13p, 18p deletion syndromes. The incidence is 1:13000, with alobar variety being the most severe form. It has a monoventricle without temporal horns, no third ventricle, no corpus callosum, no falx, a large dorsal cyst, anomalous gyri, fusion of basal ganglia, with optic nerve and pituitary hypoplasia [5]. Facial anomalies are common.

Ultrasound done at 18-20 weeks gestation is the ideal modality for screening for holoprosencephaly [6,7], though the earliest detection of the alobar variety of holoprosencephaly has been reported at 10-12 weeks using the transvaginal ultrasonography (TVS).

Conflicts of Interest

None identified

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