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Case Report

Alobar Holoprosencephaly Associated with Cebocephaly: A Case Report

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Abstract

Introduction: Holoprosencephaly occurs due to non cleavage of forebrain into two hemispheres in an embryo. It is a rare congenital disorder of brain which may be associated with facial dysmorphism. Holoprosencephaly is defect of gastrulation. It is mainly divided into three main types viz alobar, semilobar and lobar holoprosencephaly. It may be associated with midline facial defects like cebocephaly, ethmocephaly or cyclopia.

Case Report: 22 year old gravid 2 para 1 young female presented to obstetric clinical unit for general obstetric checkup. A routine sonogram for fetal well being was advised as she has not undergone any sonography during course of this pregnancy. Sonographic biometric parameters suggested gestational age of 30 weeks and 1 day. Fetal brain and face assessment revealed absent interhemispheric fissure, monoventricle, fused thalami and posterior fossa cyst in brain with reduced interorbital distance and mono nostril in face.

Discussion: Holoprosencephaly is a complex brain malformation affecting both forebrain and face. It is estimated to affect 1:16000 live births and 1: 250 conceptuses. Holoprosencephaly may be associated with midline facial anomalies like cebocephaly. Depending on the type of holoprosencephaly severity varies like alobar type are more severe followed by semilobar type.

Conclusion: Alobar holoprosencephaly has poor prognosis and may be associated with facial dysmorphism which further reduces chances of survival of fetus. Thus, early detection by prenatal ultrasound examination is important for proper management which not only help in avoiding additional burden on physical health of pregnant female but also prevents mental constrains if diagnosed early.

Keywords: Hypotelorism; Mono Nostril; Monoventricle; Holoprosencephaly

Abbreviations

HPE: Holoprosencephaly; USG: Ultrasonography

Introduction

Holoprosencephaly (HPE) is severe congenital brain malformation caused by defective fusion of cleavage of various brain structures. It is estimated to involve approximately 1:16,000 live-births. The forebrain, midbrain, and hindbrain are formed from neural tube [1]. At around 18 and 28 day of gestation the forebrain separates into two separate hemispheres but in Holoprosencephaly this separation fails to occur [2,3]. HPE is classified into three main types: alobar, semilobar and lobar. Another variant has been added to it which is known as middle interhemispheric variant (MIH) or syntelencephaly [1,4]. Alobar type is the most severe form of this anomaly. In major three types of HPE the incomplete cleavage is in the basal forebrain [1,5] whereas in the MIH variant there is fusion

of the cerebral hemispheres in the posterior frontal and parietal lobes [1,5,7]. In alobar HPE there is falx cerebri, corpus callosum and interhemispheric fissures are absent with formation of only small amount of the anterior portion of the brain [8]. Third ventricle is not present and the thalami are fused. There is partially formed cerebrum [8] and a dorsal cyst is found which correlates with thalamic fusion. This dorsal cyst is present in 92% cases of alobar HPE and less frequently with semilobar HPE (28%) and lobar HPE (9%) [1]. HPE has shown strong association with, craniofacial defects like or cleft lip, cleft palate, ocular hypotelorism (decrease distance between eyes), cyclopia (single, midline, fused eye exists in a single orbit), cebocephaly (ocular hypotelorism with a single nostril nose), single nostril, proboscis (nose-like appendage), [9]. Early detection by prenatal ultrasound examination is important for proper management and with improvements in high-resolution sonography, early diagnosis of HPE is possible.

In this present case observation sonographic diagnosis of alobar HPE with cebocephaly was done at gestational age of 30 weeks 1 day in a 22 year young women with no previous sonography record.

Case Report

A 22 year old young woman, gravida 2 and para 1, presented to the obstetrics clinical unit for normal obstetric checkup. She was having non-consanguineous marriage. She was non-diabetic, non-hypertensive having no significant medical history. Previous child was born by normal vaginal delivery with no ailment. A routine ultrasonogram was advised for general fetal well-being for fetal biometric parameters, Amniotic fluid volume and placental localisation. The sonogram revealed a singleton live intrauterine fetus with the biometric parameters suggesting gestational age of approximately 30 weeks 1 day. All biometric parameters viz Biparietal diameter (BPD), head circumference (HC), abdominal circumference (AC) and femur length (FL) were symmetrical. Sonographic evaluation of the brain revealed absence of inter hemispheric fissure, a large monoventricle (Figure 1) occupying most of area of brain, lack of sufficient cerebral matter, the two thalami were fused in mid line (Figure 2), non visualisation of cerebellar vermis and prominent cystic area in the posterior fossa. The facial details were assessed and it was observed that interorbital distance was significantly reduced suggesting hypotelorism. Assessment of nose revealed presence of a single nostril however no evidence of cleft lip was observed (Figure 3). Spine, limbs and heart did not reveal any defect. Thus, midline cranio-facial anomalies were suspected and a sonographic impression of alobar holoprosencephaly with possible cebocephaly having mono nostril was made. Gross movements of fetus and amniotic fluid volume were sufficient. Patient was referred back to the obstetric unit and followed till delivery. A normal vaginal delivery was conducted, the process of labour was uneventful and patient delivered a female baby whose general physical examination confirmed sonographic findings of hypotelorism, mono nostril i.e. cebocephaly (Figure 4 and 5).

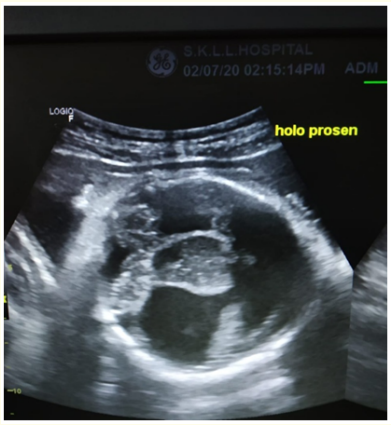

Figure 1: Sonographic image showing midline fused thalami with absent interhemispheric fissure.

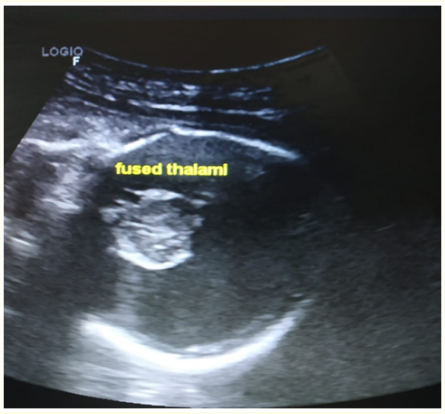

Figure 2: Sonographic image shows monoventricle with fused thalami in midline.

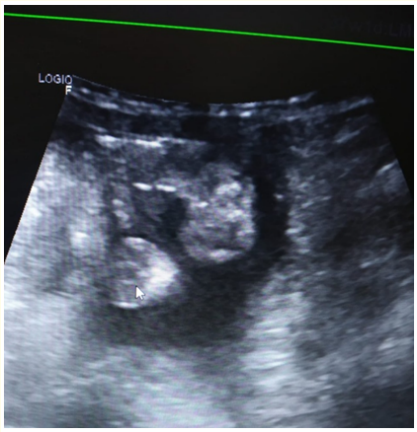

Figure 3: Sonographic image showing single nostril depicted by white arrow.

Figure 4 and 5: Post-delivery images clearly showing hypotelorism and mononostril confirming cebocephaly.

Discussion

Holoprosencephaly is a complex brain malformation affecting both forebrain and face. It is estimated to affect 1:16000 live births and 1: 250 conceptuses. Holoprosencephaly may be associated with midline facial anomalies like cebocephaly, cyclopia etc. sometimes these facial malformations may be present without cerebral malformations and are known as microforms. Prognosis of this condition is poor and if a child survives it usually have multiple medical ailments developmental delay, feeding problems, unstable heart rate and temperature, endocrine disorders like diabetes insipidus, thyroid and adrenal hypoplasia and growth hormone deficiency. The exact cause of holoprosencephaly is not known. However, it has been attributed to chromosomal abnormality like trisomy 13, maternal infections like cytomegalovirus and toxoplasmosis, environmental factors such as maternal diabetes, drugs taken during pregnancy, and low cholesterol levels [10]. Depending on the type of holoprosencephaly severity varies like alobar type are more severe followed by semilobar type. A study performed by Stashinko., *et al*. [11] involving 104 children, approximately 50% of the children with alobar HPE died within five months, and approximately 30% lived beyond one year; however, all the surviving children have multiple medical problems like they could not sit independently or speak [1,12]. In an another study conducted by Wenghoefer., *et al*. [13] on 51 children with holoprosencephaly only one child survived, however this child was having both motor and mental deficiencies. In this present case scenario there was absent corpus callosum, interhemispheric fissure, monoventricle and various facial developmental defects which fits into alobar holoprosencephaly with associated midline facial defects, cebocephaly and prognosis were very poor. This rare case shows the importance of early screening as this condition could have been easily diagnosed in late first trimester or early second trimester which could have less physical and emotional parental implications.

Conclusion

Recognising these anomalies is essential for accurate parental counseling since the prognosis is poor. There is need for careful assessment and early screening of every pregnancy for diagnoses of any anomaly and its associated outcome. If regular ultrasonography of patient is done during pregnancy early diagnosis of severe congenital conditions like these can be done which will have less physical and emotional implications.

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