

Antenatal Sonographic Diagnosis of a Case of Alobar Holoprosencephaly: A Case Report

Rizwan Ahmad Khan¹, Manjari Thapa², Shagufta Wahab^{2*}

¹Division of Pediatric Surgery, Department of Surgery, JN Medical College, Aligarh Muslim University, Aligarh, India; ²Department of Radiodiagnosis, JN Medical College, Aligarh Muslim University, Aligarh, India.
Email: *drshaguftawahab@rediffmail.com

Received May 2nd, 2012; revised June 6th, 2012; accepted July 19th, 2012

ABSTRACT

Holoprosencephaly is a rare congenital brain malformation resulting from failure of diverticulation and cleavage of primitive prosencephalon which occurs at 4th - 8th week of gestation and is usually associated with multiple midline facial anomalies. Herein we report an antenatal case of such patient. Patient was evaluated and because of the magnitude of the problem induction was done which led to a still born baby.

Keywords: Holoprosencephaly; Alobar; Antenatal Detection; Developing Countries; Prognosis

1. Introduction

Holoprosencephaly is a rare congenital brain malformation resulting from failure of diverticulation and cleavage of primitive prosencephalon which occurs at 4th - 8th week of gestation and is usually associated with multiple midline facial anomalies. This disorder is either incompatible with life or infants suffer from varying grade of mental retardation.

Sonography is an excellent non-invasive tool for prenatal diagnosis of holoprosencephaly. This case report describes a case of alobar holoprosencephaly diagnosed prenatally with ultrasound and highlights the importance of high-resolution real time imaging. We want to stress that the condition is rare and therefore lack of familiarity may lead to diagnostic confusion.

2. Case Report

A 24-year-old, gravida 2 and para 1 underwent antenatal ultrasonography at 24 menstrual weeks as determined by dates, which disclosed increased intracranial fluid in the fetus. Past medical history and family history were unremarkable, with the previous child being born at term with no congenital anomaly. A repeat ultrasound examination demonstrated a monoventricle fluid-filled cavity. The cerebral cortex was represented by thinned out mantle. Two orbits were seen; no clear evidence of proboscis was seen. The midbrain and posterior fossa appeared normal (**Figures 1**). An increased amount of amniotic fluid (polyhydramnios) was present. Based on the ante-

natal sonographic findings a diagnosis of alobar holoprosencephaly was made. Preterm induction of vaginal delivery resulted in a stillborn fetus. Examination of the fetus demonstrated only proptosis but no gross facial anomaly. Brain morphology could not be assessed due to autolysis. Chromosomal analysis disclosed a trisomy 13 karyotype. Patient was investigated as per the guidelines set by the Ethics committee of our hospital.

3. Discussion

The incidence of holoprosencephaly is approximately 1 - 1.4 per 10,000 live births [1] but as the incidence of



Figure 1. Antenatal sonogram of fetal head (coronal view) with alobar holoprosencephaly showing monoventricular cavity with surrounding thinned cerebral tissue, fused thalami and absence of midline structures such as falx cerebri. Axial view of the same patient showing fused thalami and thinned out cortical mantle of the fetal head.

*Corresponding author.

spontaneous abortions is high, the actual incidence may be quite high. Both environmental factors and genetics are suspected in its etiology and approximately 30% of the cases are associated with chromosomal defects mostly trisomy 13 and 18. However most of the cases have a normal karyotype [2]. It represents a spectrum of disorder with most severe form known as alobar type showing absolutely no cleavage of prosencephalon with a large monoventricular system surrounded by a thinned out mantle with absent falx cerebri, corpus callosum, fornix, optic tract and olfactory bulbs. The thalami are fused with absent third ventricle and the single ventricle usually communicates with a dorsal sac [3]. The infants are stillborn or survive only upto few days or weeks. Semilobar holoprosencephaly is an intermediate form with posterior partial separation of two hemispheres and ventricles thus a rudimentary occipital horn with a small portion of falx and interhemispheric fissure in the occipital cortex may be seen. Thalami are partially separated with a rudimentary third ventricle [3]. The least severe form is the lobar variety showing almost complete separation of cerebral hemispheres and lateral ventricle with developed falx and interhemispheric fissure so prenatal diagnosis may be difficult. Cavum septum pellucidum is however absent and there is fusion of frontal horns [3]. This syndrome is associated with mild mental retardation. The mildest form is septo-optic dysplasia with absent septum pellucidum, optic nerve hypoplasia and associated hypothalamic-pituitary dysfunction. As the face develops at the same time as the diverticulation of brain, holoprosencephaly is accompanied by a number of midline facial anomalies and the degree of facial dysmorphism parallels the severity of holoprosencephaly so sonographic evaluation of facial morphology may help in prenatal diagnosis of holoprosencephaly. Studying the strong association between facial anomalies and holoprosencephaly, DeMeyer commented "brain malformations can be predicted by patient's face" [4]. The severest facial malformation like cyclopia, ethmocephaly and cebocephaly occurred only with alobar holoprosencephaly [5] with semilobar and lobar types showing milder facial anomalies. These are hypotelorism, median cleft lip and palate, lateral cleft lip and palate and pyriform aperture stenosis [5-7]. However the face may even be almost normal in few cases of alobar holoprosencephaly as in our case. Other extrafacial anomalies are also common with holoprosencephaly and includes polydactyly, renal dysplasia, omphalocele, club foot, esophageal atresia, cardiac malformation etc. [4,5]. Routine obstetric USG is performed during the first trimester preferably between 11th - 14th week of gestation and besides determining the gestational age, number of fetuses and presence of any complication, USG can also detect a number of con-

genital anomalies. Sonography is most helpful in the prenatal diagnosis of holoprosencephaly especially of alobar type and is the decisive modality for the management and follow-up of such cases so that the mother can opt for termination of pregnancy and the doctor can decide for a vaginal delivery rather than caesarean section [8-10].

4. Conclusion

Although rare, facial and brain anomalies can be recognized in high resolution ultrasonography probe is used. Interorbital spaces, ventricular configuration and facial morphology should be looked for in both the planes.

REFERENCES

- [1] M. Epelman, A. Daneman, S. I. Blaser, C. Ortiz-Neira, O. Konen, J. Jarrin and O. M. Navarro, "Differential Diagnosis of Intracranial Cystic Lesions at Head US: Correlation with CT and MR Imaging," *Radiographics*, Vol. 26, 2006, pp. 173-196. doi:10.1148/rg.261055033
- [2] R. A. Filly, D. H. Chinn and P. W. Callen, "Alobar Holoprosencephaly: Ultrasonographic Prenatal Diagnosis," *Radiology*, Vol. 151, No. 2, 1984, pp. 455-459.
- [3] C. Manelfe and A. Sevely, "Neuroradiological Study of Holoprosencephalies," *Journal of Neuroradiology*, Vol. 9, No. 1, 1982, pp. 15-45.
- [4] W. DeMyer, W. Zeman and C. Palmer, "The Face Predicts the Brain: Diagnostic Significance of Median Facial Anomalies for Holoprosencephaly (Arhinencephaly)," *Pediatrics*, Vol. 34, No. 2, 1964, pp. 256-263.
- [5] J. P. McGahan, D. A. Nyberg and L. A. Mack, "Sonography of Facial Features of Alobar and Semilobar Holoprosencephaly," *American Journal of Roentgenology*, Vol. 154, No. 1, 1990, pp. 143-148.
- [6] J. R. Mernagh, P. T. Mohide, R. E. Lappalainen and J. G. Fedoryshin, "US Assessment of the Fetal Head and Neck: A State of the Art Pictorial Review," *Radiographics*, Vol. 19, 1999, pp. S229-S241.
- [7] K. K. W. Fong, A. Toi, S. Salem, L. K. Hornberger, D. Chitayat, S. J. Keating, F. McAuliffe and J. A. Johnson, "Detection of Fetal Structural Abnormalities with US during Early Pregnancy," *Radiographics*, Vol. 24, No. 1, 2004, pp. 157-174.
- [8] D. A. Nyberg, L. A. Mack, A. Bronstein, J. Hirsch and R. A. Pagon, "Holoprosencephaly: Prenatal Sonographic diagnosis," *American Journal of Roentgenology*, Vol. 149, No. 5, 1987, pp. 1051-1058.
- [9] L. H. Lowe, T. N. Booth, J. M. Joglar and N. K. Rollins, "Midface Anomalies in Children," *Radiographics*, Vol. 20, No. 5, 2000, pp. 907-922.
- [10] P. D. Cayea, I. Balcar, O. Alberti Jr. and T. B. Jones, "Prenatal Diagnosis of Semilobar Holoprosencephaly," *American Journal of Roentgenology*, Vol. 142, No. 2, 1984, pp. 401-402.