ORIGINAL RESEARCH PAPER
Volume - 10 | Issue - 03 | March - 2021 | PRINT ISSN No. 2277 - 8179 | DOI : 10.36106/ijsr
INTERNATIONAL JOURNAL OF SCIENTIFIC RESEARCH

ALOBAR HOLOPROSENCEPHALY DIAGNOSED IN AN ANTENATAL PATIENT : A CASE REPORT

Radiodiagnosis

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ABSTRACT

Holoprosencephaly is the congenital cerebrofacial anomaly of variable severity. I report a case of alobar type of holoprosencephaly in a twenty one years old primigravida who presented for routine sonography. The sonographic scan revealed a single live intrauterine fetus at nineteen weeks of gestation with absent falx, fused thalami, monoventricle, proboscis, and cyclopia. Fetal MRI was performed which confirmed the sonographic findings. The aim of presenting this classical case of alobar holoprosencephaly is to sensitize the clinicians and radiologists to the imaging features of holoprosencephaly and to stress the importance of early diagnosis. If this condition is diagnosed at an early period of gestation then termination can be done which may reduced the psychological trauma to the mother for bearing a deformed fetus.

KEYWORDS

Holoprosencephaly, alobar, monoventricle cyclopia, proboscis.

INTRODUCTION

Holosprosencephaly is the structural malformation of the brain characterized by abnormal cleavage of the prosencephalon during embryonic development. The prosencephalon leads to the formation of cerebral hemispheres,thalami and the basal ganglia. Therefore, the abnormal development of the prosencephalon results in variable fusion anomalies of these structures.The alobar type of holoprosencephaly is the most severe form which is incompatible with the life [1]. So, early diagnosis of this clinical entity allows for early termination of pregnancy.

Case History

21 years old primigravida presented to the department of radiodiagnosis at IGMC, Shimla for routine sonographic scan. On clinical examination, approximate fundal height was corresponding to 18 to 20 weeks. The clinical and biochemical examination was within normal limits. There was no history of consanguinity of marriage.

On sonography there was single live intrauterine fetus with an average gestational age corresponding to 19 weeks. The fetal brain appeared deformed with loss of normal ventricular system. There was a single large central ventricle, with fused thalami in between (Figure 1a). The falx cerebri and septum pellucidum were not visualized. There was an external median tubular soft tissue projection suggestive of proboscis seen in the face, at the level of the frontal bones without intracranial extension (Figure 1b). The facial structures were also appearing dysmorphic with fused orbits. The rest of the major organs including spine, thoracic cage, heart, and limbs were sonologically normal. Amniotic fluid was adequate. For further evaluation fetal MRI was performed on 1.5T MRI machine that confirmed the sonographic diagnosis (Figures 2a and 2b). There was no evidence of any other associated anomaly on MRI. The medical termination of pregnancy was done which revealed all the findings observed on imaging. No additional dysmorphic features seen.

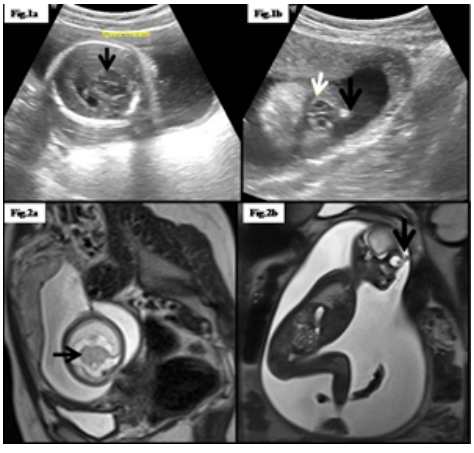

Fig. 1a & 2a show the fused bilateral thalami (black arrow) with single monoventricle while white arrow in fig.1b shows fused bilateral orbits and black arrow in fig.1b&2b shows the proboscis.

DISCUSSION

Holoprosencephaly constitutes a spectrum of cerebrofacial anomalies which occurs due to complete or partial failure of the cleavage of the primitive forebrain [1,2]. During the 4th week of the gestation, the neural tube leads to the formation of three segments of the primitive brain, namely, prosencephalon, mesencephalon, and rhombencephalon. By the 5th week of gestation, the prosencephalon further divides into the telencephalon and diencephalon. Later, the telencephalon forms the two cerebral hemispheres whereas the diencephalon forms the thalami, the hypothalamus, and the basal ganglia. The midline facial structures develop from the prechordal mesoderm. The severity of the intracranial abnormalities is in proportion to degree of facial dysmorphism. Therefore any of the dysmorphic facial features should direct the sonologist to search for the CNS anomalies. This observation has led to the popular statement "face predicts the brain" by DeMeyer [3]. Cyclopia is the most severe [4] whereas mild hypotelorism with flat face is the least severe abnormality amongst the median cerebrofacial anomalies [5]. Upto 58% of the facial abnormalities can be detected by sonography [5]. In my case the two classical facial anomalies were seen, namely, cyclopia in the which there is absence of the normal eye balls and a median horizontally placed eye in the fore head, and proboscis which is the median primitive non-canalized nose as a projection from the forehead. The forebrain is responsible for induction of proper development of the orbits during the embryonic development [6]. Thus, improper diverticulation and cleavage of the forebrain results in improper induction of the formation of orbits and further leads to cyclopia as seen in this case [6]. There are three main subtypes of holoprosencephaly, namely, alobar, semilobar, and lobar types. The alobar holoprosencephaly is the most severe form and shows undifferentiated holosphere of the cerebral parenchyma with a central enlarged monoventricle and fused thalami [2]. The falx, interhemispheric fissure, corpus callosum, optic tracts, olfactory bulbs, and the septum pellucidum are also absent [1]. A dorsal cyst may also be seen in the posterior cranial fossa in very severe forms of holoprosencephaly [1]. The semilobar variety is of intermediate severity with early midline differentiation and sagittal separation [7]. It shows a rudimentary falx, partial interhemispheric fissure, absent septum pellucidum, partial separation of thalami, and large H-shaped monoventricle with variable fusion of basal ganglia. The lobar type is the mildest among the three characterized by near total cleavage of cerebral hemispheres, presence of falx, interhemispheric fissure, and absent septum pellucidum [8]. The frontal horns also appear squared off or box like due to the absence of septum pellucidum. The thalami and the basal ganglia are separated and normal in this variety. The alobar form of holoprosencephaly is incompatible with life whereas children with semilobar and lobar variants have variable survival. The role of fetal MRI is to confirm the sonographic findings and to detect any other associated anomaly. The diagnosis of holoprosencephaly before 20 weeks of gestation by imaging is important in order to avoid the psychological stress to the mother of bearing the deformed fetus.

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