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Holoprosencephaly Alobar: Imaging Finding: A Case Report and Literature Review

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Abstract **Case Report**

Holoprosencephaly is a rare and a complex congenital brain malformation resulting from defective cleavage of the prosencephalon that occurs at 4 - 8th week of gestation and is usually associated with multiple cranio facial anomalies. It is the most common forebrain developmental anomaly in humans and a worldwide distribution. The etiology of HPE is very heterogeneous it has an extremely reserved fetal prognosis, particularly for the alobar form. Here, we report a case of holoprosencephaly alobar of a 24-day old boy, with microcephaly, flat nose, a single nostril, midline cleft lip palate and choanal atresia.

Keywords: Holoprosencephaly alobar, imaging CT.

INTRODUCTION

Holoprosencephaly (HPE) is a rare congenital anomaly, it results from an anomaly in the division of the forebrain into cerebral hemispheres, during the second month of gestation [1]. It includes three classic forms alobar, semi-lobar and lobar HPE. The alobar form is the most severe, and results from a lack of longitudinal clivage of the hemispheres. It can be the cause of facial abnormalities, neurological manifestations and various endocrine disorders [2]. We report two cases of alobar holoprosencephaly associated with multiple facial deformities.

CASE REPORT

It was a 30 day old male infant, the first pregnancy of a first degree consanguineous parents. No obstetric ultrasound was performed during pregnancy. After vaginal delivery, a transfontanellar echography and cerebral CT scan were performed in our department, which revealed a single ventricle, extending posteriorly to the occipital vault, while the brain tissue in front of this single ventricle has a "horseshoe" appearance, absence of the callosal commissure (corpus callosum), absence of bulbo-peduncular and cerebellar hemispheres morphological abnormalities. A fourth ventricle with a normal appearance and an arterial circle of the base of the brain with no abnormality. A single nostril, choanal atresia, a median cleft palate and a solitary median maxillary central incisor were reported. The abnormalities of the supratensorial structures are characteristic and have enabled us to retain the diagnosis of alobar HPE associated with facial malformation (Figures 1). The newborn was referred to the neurosurgery department where he was followed up but the evolution was marked by respiratory instability and the newborn died.

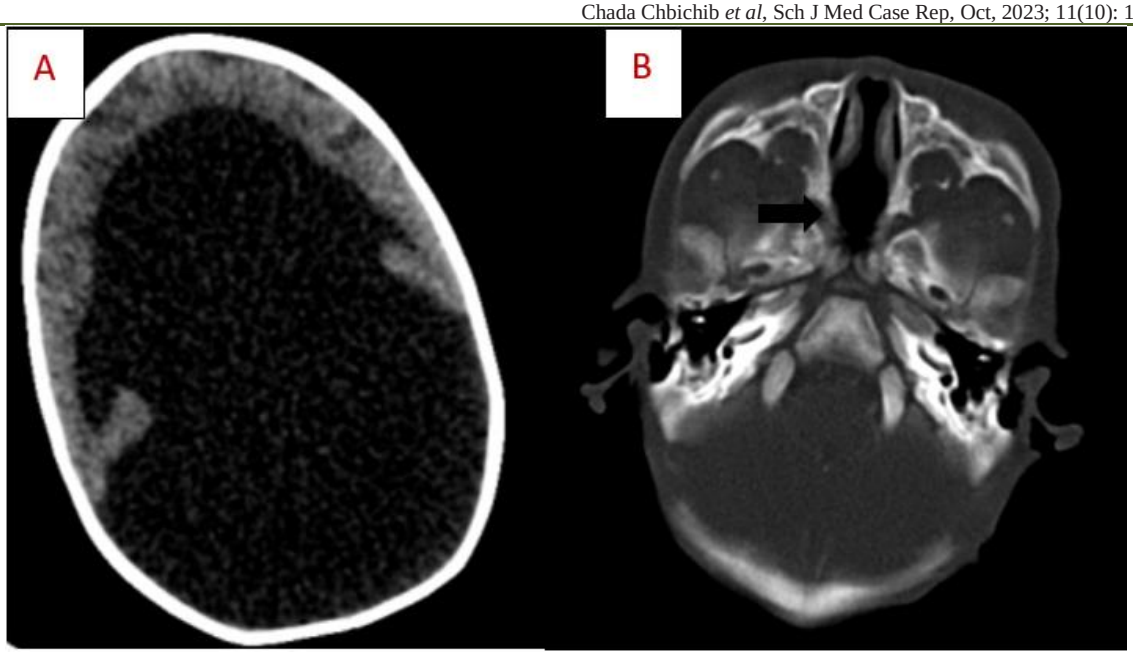

Figure 1: A: Axial CT images showing a single large U-shaped ventricular cavity, fused thalamus with a thin rim of peripheral cerebral parenchyma and an absence of the interhemispheric fissure. B: A single nostril and choanal atresia (black arrow)

DISCUSSION

HPE is a potentially serious malformative anomaly of the central nervous system related to a defect in the formation and individualization of the cerebral hemispheres. It results from a cleavage anomaly of the prosencephalon during the second month of pregnancy. HPE results from a defect induction of neurectoderm by the prechordal plate during the third week of embryonic life. The result is an anomaly in the development of the prosencephalon, consisting of a lack of evagination of the prosencephalic vesicles. As a result, a hemispheric mass is formed, replacing the two cerebral hemispheres, with the absence of medial structures, notably the commissures. It is also associated with structures derived from the prosencephalic vesicles and the diencephalic vesicle. The epidemiology of holoprosencephaly is inadequately described, in part because the most benign forms may go undetected; on the other side, there is a significant natural loss of the fetus. As a result, the prevalence rate differs between study groups [5]. Genetic factors and environmental agents dominate the etiologies of HPE. The genetic etiopathogenesis of HPE is widely accepted, and is dominated by chromosome 13 abnormalities. HPE is usually associated with different types of malformations visceral and/or skeletal malformations. Isolated, non-sporadic familial HPE, with normal caryotype, has an autosomal dominant mode of transmission, less frequently autosomal recessive or X-linked. Currently, four genes involved in the development of HPE have been identified: mainly the sonic hedgehog (Shh) gene. Some environmental and/or maternal teratogenic factors, in particular maternal diabetes, maternal alcohol consumption, retinoids and congenital cytomegalovirus infection [6]. In the presented case, no risk factors were identified in the mother's history. No genetic studies were made on the infant. There are classically three anatomical varieties of HPE of decreasing severity:

1. Alobar, being the most severe form and characterized by undifferentiated holosphere of the cerebral parenchyma with a central monoventricle, fused thalami, and absence of midline structures, such as corpus callosum and the midline falx cerebri, and facial dysmorphism which include such abnormalities as cyclopia, proboscis, ethmocephaly and cebocephaly. It is the most severe form.
2. Semilobar, is characterized by the presence of a median line of variable length that forms at the posterior part of the holosphere an outline of two occipital lobes.The olfactory lobes are absent. microcephaly is usually observed.
3. Lobar, is the minor form, characterized by an entirely developed cerebral hemispheres, distinct interhemispheric sulcus, continuous frontal neocortex on the midline, absence of the callosal commissure, hypoplastic or normal separation of the basal ganglia.

Another milder subtype of HPE, called middle interhemispheric variant (MIHF) or syntelencephaly, has been recognized. characterized by the absence of separation of the posterior portions of the frontal and parietal lobes, genu callosal and splenium normally formed. Absence of hypothalamus and lentiform nuclei normally separated [7]. The semiology of this affection can be grouped into three syndromes:

- A dysmorphic syndrome, frequently encountered in alobar holoprosencephaly with cyclopia, ethmocephalia, cebocephaly, median or bilateral cleft lip or bilateral cleft lip or hypertelorism. This syndrome is not constant,

- A neurological syndrome with major disorders as early as the neonatal period (apneas and tonic spasms with, on examination axial hypotonia and spasticity of the limbs) or psychomotor retardation,
- An endocrine syndrome such as ante- or post-hypophyseal insufficiency [6-7].

Antenatal diagnosis of HPE is based on conventional obstetric ultrasound supplemented by fetal MRI, showing cerebral anomalies such as a single ventricular cavity with absence or abnormal development of medial structures, thalami fusion and microcephaly. These anomalies are more or less depending on the anatomical form of HPE. Ultrasound diagnosis of HPE can be made early in the first trimester of pregnancy, enabling early interruption of pregnancy, in particular in the case of alobar and semi-lobar forms. In lobar cases diagnosis could be difficult because the antenatal picture of septo-optic dysplasia is almost identical to that of lobar holoprosencephaly [8, 9]. In our case, the pregnancy was not observed, the diagnosis is made by transfontanel ultrasound, which will be confirmed by a CT scan. An angiographic study can also be contributory [10]. The management of EPH remains complex and difficult, and can only be performed by a multidisciplinary team [10]. Treatment may include medication (hormone replacement, antiepileptics, feeding tube, rehabilitation medicine, close monitoring) and/or surgery (gastrostomy tube, cleft lip and palate repair, ventriculoperitoneal shunt). Whatever the management modalities, HPE in these anatomical varieties remains a pathology with a poor prognosis. [6].

CONCLUSION

HPE is a rare congenital brain malformation, frequently associated with facial anomalies. Its prognosis is often poor, particularly for the alobar form. The etiologies of HPE are multiple, dominated by genetic and environmental anomalies mainly represented by maternal diabetes. The prenatal diagnosis of HPE is possible before the third gestational month, when suggestive images are detected on obstetrical ultrasound and magnetic resonance imaging. Such early diagnosis allows early management adapted to the condition.

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