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Case Report

Holoprosencephaly Alobar: Contribution of Cerebral CT Scan in a Case Report

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Abstract

Holoprosencephaly is a rare brain anomaly that results from incomplete cleavage of the primitive forebrain prosencephalon during early embryogenesis. It includes several rare complex and heterogeneous disorders. The alobar form is associated with an unfavorable fetal prognosis. The aim of this observation is to review the different aspects of holoprosencephaly through a case of alobar holoprosencephaly diagnosed on a cerebral CT scan in the presence of congenital hydrocephalus without facial abnormalities of a 12-day-old female newborn.

Keywords: *Alobar Holoprosencephaly; Hydrocephalus; CT Scan*

Introduction

Holoprosencephaly is a complicated congenital brain malformation, that occurs between days 18th and 28th day of gestation [1]. It results from an incomplete division of the prosencephalon and affects both the brain and the face [1,2]. It occurs in up to 1 of 250 pregnancies, but only in 1 of 8000 to 1 of 10,000 live births [2- 4]. Holoprosencephaly is classified into 4 subtypes: alobar, semi-lobar, lobar, and the middle interhemispheric. The most severe subtype is the alobar form, which is characterized by a complete absence of division of the primitive brain into two hemispheres [4-6]. The aim of this observation is to review the different aspects of Holoprosencephaly through a case of alobar holoprosencephaly diagnosed on a cerebral CT scan in the presence of congenital hydrocephalus without facial abnormalities of a 12-day-old female newborn.

Case Report

A 12-day-old female newborn, first child's couple, has been referred to the radiology department for cerebral CT scan so as to investigate a hydrocephalus with congenital moderate macrocephaly. It is a question of a newborn from a poorly monitored pregnancy with a third trimester ultrasound suggestive of hydrocephalus. The mother, aged 24 years, had no known pathological history or chronic pathology. There was no notion of consanguinity. The investigation did not find any teratogenic drugs taken during pregnancy. Clinically, there was no facial malformation, the sucking reflex was present but automatic walking and grasping were impossible; the head circumference was 30 cm (+2 SD); the weight was 48 cm (-2 SD) and the weight 2.7 kg (-1 SD).

The cerebral scan performed without enhancement (Figure 1) confirmed the encephalic malformation by showing alobar holoprosencephaly, which was manifested by a single dilated ventricular cavity pushing forward the remaining cerebral parenchyma giving a "horseshoe" appearance, associated with a fusion of the thalami and an absence of the interhemispheric scissure and the callosal commissure (corpus callosum). The fourth ventricle had a normal appearance. The abnormalities of the supratentorial structures are characteristic and have enabled us to retain the diagnosis of alobar holoprosencephaly without associated facial malformation. The newborn was referred to the neurosurgery department where she was followed up: monitoring of the hydrocephalus and the cranial perimeter which were stable and did not yet require a bypass, a thyroid check-up and an ionogram which were normal.

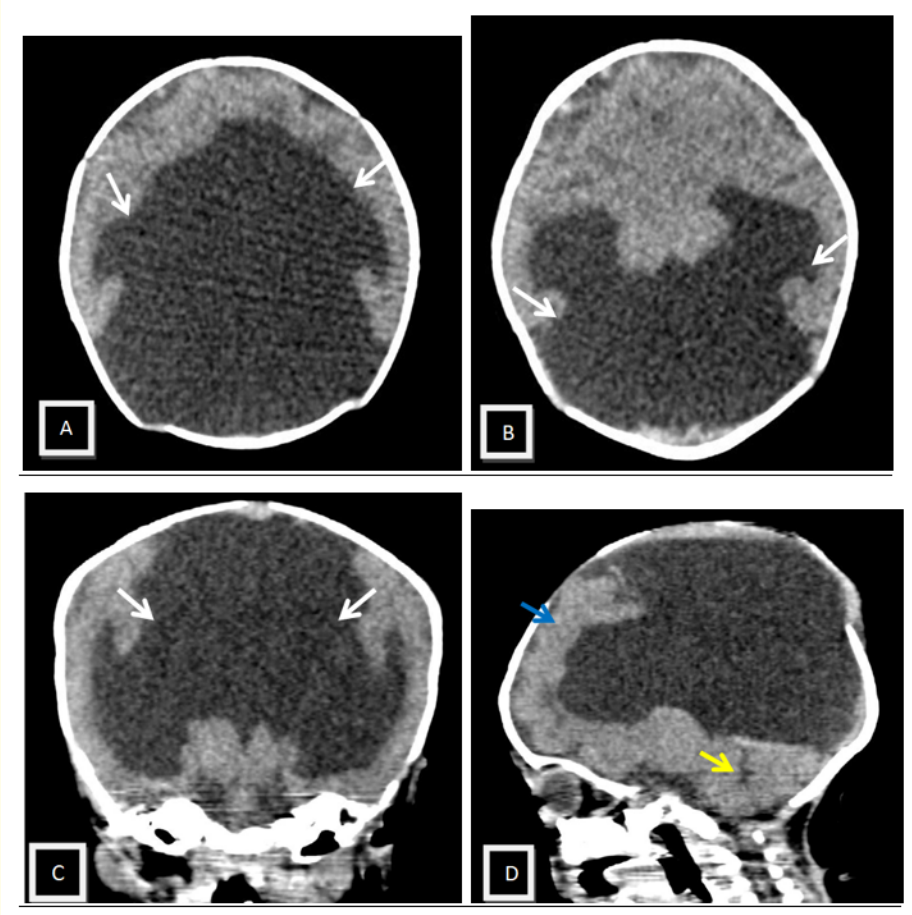

Figure 1: *Cerebral CT scan, axial section (A, B), coronal reformatted scan(C) and sagittal reformatted scan (D), displaying a single dilated supratentorial cavity (white arrow) pushing forward the remaining cerebral parenchyma giving a "horseshoe" appearance (blue arrow). The fourth ventricle had a normal appearance (yellow arrow).*

Discussion

Alobar holoprosencephaly is the most severe of the classical forms of holoprosencephaly [1]. Various etiologies have been reported, including chromosomal abnormalities (trisomy 13, 18) [5], polymalformative syndromes (CHARGE syndrome) [5], and environmental

factors (diabetes or hypocholesterolemia during pregnancy) [1]. The disorder is often associated with craniofacial abnormalities [4,6], given the common origin of the embryonic forebrain and midface from the prechordal mesoderm [7]. The most common craniofacial anomalies reported in the literature are micro-crania, hypotelorism, and median cleft lip/palate [4]. Sometimes normal facial appearance can be observed [4,8]. In our case, there were no craniofacial malformations. Other anomalies of the median lineage can be observed. One case of oropharyngeal stenosis has been reported [4].

The diagnosis of holoprosencephaly is based above all on imaging, which also allows the establishment of a prognosis classification by identifying the 4 subtypes. The diagnosis is primarily based on ultrasound and MRI during the third trimester of pregnancy, transfontanellar ultrasound, CT scan, and particularly brain MRI on postnatal [8,9], and genetic diagnosis, which is based on karyotype to search for a chromosomal anomaly in the newborn (trisomy 13 or 18 or triploidy), but also the parents, because if one of them is carried a balanced translocation, the risk of recurrence is high [10].

Alobar holoprosencephaly: is the most severe and often non-viable form, in which the cleavage defect is complete. The brain is small and contains a single ventricular cavity (representing the third ventricle and lateral ventricles). A fluid pocket called a "dorsal sac", of variable size, communicates widely with the ventricular cavity. Facial malformations are usually more complex. The cerebral CT scan findings are characteristic by showing a large single ventricular cavity, limited behind by a membrane and bordered in front of the remaining cerebral parenchyma, realizing the "horseshoe" appearance. The third ventricle is not individualized, and the basal ganglia are fused on the median line. At the sub-tensor level, the brainstem, the fourth ventricle, and the cerebellum is present and can be normal, hypoplastic, or malformed (cyst). Our patient presented a stage of alobar holoprosencephaly with dilatation of a ventricular cavity and a fusion of the thalami and agenesis of the corpus callosum.

Semi-lobar holoprosencephaly: This is an intermediate form in which there is a median sulcus but only in the posterior part of the hemisphere with an outline of the two occipital lobes.

Lobar holoprosencephaly: this is the least severe form and the lobes of the brain lobes appear well developed and a median sulcus seems to separate them. In reality on coronal MRI sections, there is no true interhemispheric cleft and there is a single ventricle without a medial structure.

The middle interhemispheric: The least common type of HPE. It's a defect of separation of the posterior portions of frontal lobes and the parietal lobes, with varying lack of cleavage of the basal ganglia and thalami and absence of the body of the corpus callosum but the presence of the genu and splenium of the corpus callosum [7].

Antenatal ultrasound evaluation (transabdominal or transvaginal) can reliably identify most cases of alobar and semi-lobar holoprosencephaly but has poorer accuracy in diagnosing lobar holoprosencephaly [6].

MRI gives precise information on the medial structures and allows the identification of the most discrete abnormalities [10].

The management of children with holoprosencephaly requires multidisciplinary care, which aims to detect the complications to avoid further disability and improve their quality of life [8].

Conclusion

Holoprosencephaly is a complex brain malformation, affecting both the brain and the face, resulting from an incomplete division of the prosencephalon. Imaging plays an important role in the diagnosis of this entity in the antenatal or postnatal period and relies on ultrasound, CT scan, and MRI.

The approach to therapeutic management is multidisciplinary, and the treatment remains symptomatic palliative. In the majority of cases, the prognosis is reserved.

Conflict of Interest

The author(s) declared no potential conflicts of interest with respect to the research, authorship, and/or publication of this article.

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