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Alobar Holoprosencephaly in a Neonate: A Rare Case Report and Review of the Literature

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Abstract

Holoprosencephaly (HPE) is a rare brain malformation with multiple etiologies and is often associated with suggestive facial anomalies. This pathology is the result of a defect in the early development of the forebrain. There are three clinical forms: lobar, semi-lobar, alobar and another milder subtype of HPE called middle interhemispheric. In this clinical case, we present a newborn with alobar holoprosencephaly and we highlight the clinical, radiological and progressive clinical aspects of this illness during the neonatal period.

Keywords

Polymalformative Syndrome, Holoprosencephaly, Hydrocephalus

1. Introduction

Holoprosencephaly is a rare cerebral malformation secondary to abnormal cleavage of the forebrain and is characterized by the formation of a midline hemispheric mass [1]. The life expectancy of individuals with HPE varies and is dependent on the severity of the defect [2]. HPE can also be a component of several genetic syndromes, and in many cases, the cause is unknown [2].

The most severe type of HPE, known as alobar HPE, is life-threatening and manifests clinically as facial dysmorphism with hypotelorism, microcephaly, and a blind ended nostril [3].

In order to increase awareness of the value of early prenatal detection and counseling, we will present a case study of alobar HPE with literature review.

2. Clinical Observation

We report the case of a newborn female from a non-consanguineous marriage and a poorly followed pregnancy because of low socioeconomic status and lack

of knowledge about the advantages of prenatal care. The pregnancy was estimated at term, medicalized by vaginal delivery. The mother, aged 38 years, had no known pathological history or chronic pathology. The clinical examination on admission revealed: signs of struggle with a Silverman score of 2/10, severe dehydration, a facial dysmorphia (Figure 1), with a single nostril opening, agenesis of the nasal septum, a large median cleft lip and hypotelorism.

There were no other obvious malformations on the rest of the clinical examination. Chest X-ray showed apical opacities probably by false route due to the anomaly of the palate, cardiac and abdominal ultrasound showed no malformation. Trans fontanellar ultrasound revealed significant hydrocephalus and a complementary brain magnetic resonance imaging MRI (Figure 2(a), Figure 2(b)) showed: alobar holoprosencephaly, agenesis of the corpus callosum, a single ventricular cavity with hydrocephalus, posterior supra tentorial cyst formation and right globe hypotrophy.

Biologically, he had renal insufficiency which subsequently corrected. During follow-up, a severe and sustained hypernatremia of the order of 165 meq /L was noted.

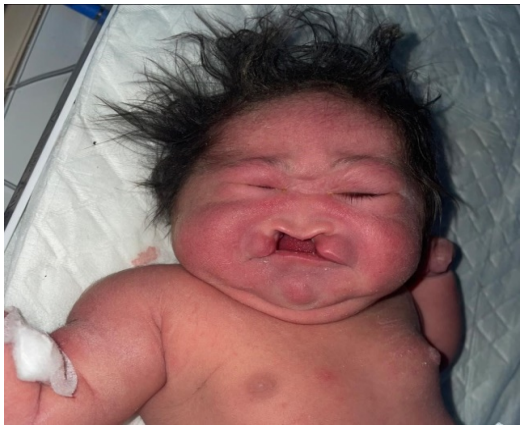
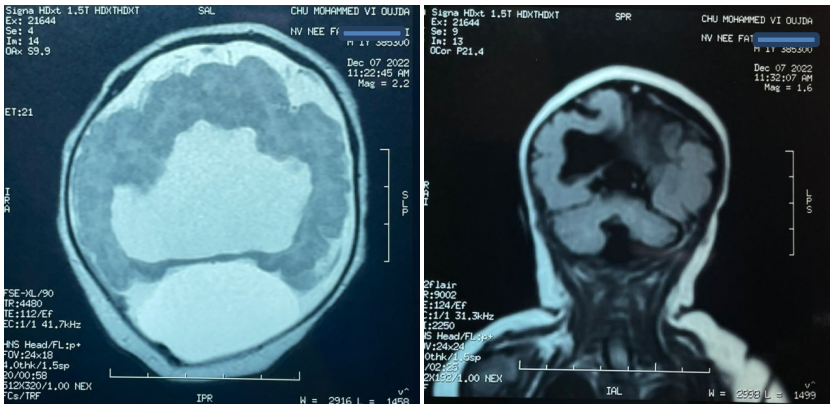


Figure 1. Facial dysmorphia of the hypotelorism type and median cleft lip and palate.



(a) (b)

Figure 2. (a & b) MRI axial and coronal sections showing: dilation of a single ventricular cavity with absence of inter hemispheric fissure.

In addition, a genetic study was performed to search for mutations in the genes NOTCH [inter-cellular signaling], FGF [fibroblastic growth factor], NODAL [member of the transforming growth factor b family], SHH [sonic hedgehog] and the newborn's karyotype did not reveal any abnormality.

Diabetes insipidus and any etiology of hypernatremia were ruled out, the diagnosis of neurogenic hypernatremia was maintained and good hydration with follow-up was recommended. Research was conducted to look for endocrine abnormalities, specifically, an 8-hour cortisol and a thyroid check-up check was done, and the results showed no abnormalities. The evolution was characterized by the installation of an opisthotonos posture (**Figure 3**) despite motor physiotherapy.

Figure 3. The result of alobar holoprosencephaly on day 40 of life: disposition in opisthotonos.

3. Discussion

Holoprosencephaly (HPE) is a severe and complex congenital brain malformation associated with specific facial abnormalities. The abortifacient nature of the malformation and the minor forms that may go unnoticed contribute to the estimated prevalence of less than 4.4 in 10,000 live births [4]. Several risk factors, including consanguinity and the incrimination of various genetic mutations, have been reported in the literature. The majority of authors note a female predominance and maternal age over 30 years, which is consistent with our case. According to the degree of individualization of the cerebral hemispheres, Demyer and Zeman [5] described three anatomical forms of HPE: alobar, semi-lobar and lobar.

The first two forms have a very poor prognosis, justifying early medical termination of pregnancy. For alobar HPE is which remains the most severe form, in which the telencephalon consists of a holosphere containing a single ventricular cavity closed in its posterior part by a thin wall, giving it a pseudo cystic appearance, Microcephaly is present consistently, the olfactory lobes are absent [5]. Several types of central nervous system malformations may be associated with Holoprosencephaly such as atresia of the aqueduct of Sylvius, cerebellar

hypoplasia, vermiform agenesis, Dandy-Walker syndrome and neural tube closure anomalies.

Ocular anomalies occur in one quarter to one third of cases: the development of the eye blanks begins in the fourth week in the forebrain, near the future diencephalon. In addition to single eye and synophthalmos, microphthalmos, cataract, retinal dysplasia and retinal coloboma may be observed [6].

Neurogenic hypernatremia is a rare complication holoprosencephaly, related to a hypothalamic damage secondary to changes in the midline defining holoprosencephaly, must be clearly distinguished from others causes of hypernatremia [7].

Prenatal diagnosis of HPE is possible just by fetal ultrasound. It is based on the association between intracranial symptoms and facial anomalies, particularly in the complete form [4]. While alobar and semi-lobar forms of HPE can be identified in the first trimester [4]. Prenatal diagnosis was not performed on this patient because the pregnancy was poorly monitored. It was made postnatally by imaging (brain MRI).

The prognosis is often poor, even leading to death [6]. In the case of our patient (currently 3 months), the evolution was marked by motor and postural anomalies with an opisthotonos attitude and a persistent hypernatremia of neurological origin related to a midline anomaly.

In front of the first birth of a child with HPE, genetic counseling is necessary to evaluate the risk of recurrence and the importance of prevention without ignoring the immense interest of a follow-up of the subsequent pregnancies.

4. Conclusion

A rare cerebral malformation known as holoprosencephaly was once thought to be fatal, but today's prognosis depends on the type of HPE and associated facial anomalies. The best diagnostic procedure is an ultrasound examination followed by an MRI. The prognosis is reserved, hence the need for a good follow-up of pregnancies to make the antenatal diagnosis and take the necessary measures, although the discussion of therapeutic termination of pregnancy still remains a dilemma in our tale.

Consent

Written informed consent was obtained from the patient parents for publication of this case report and accompanying images.

Author Contributions

All authors contributed to the conduct of this work. All authors also declare that they have read and approved the final version of the manuscript.

Conflicts of Interest

The authors declare no conflict of interest.

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