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Case Report

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Alobar Holoprosencephaly: About A Case

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Abstract

Holoprosencephaly is a rare cerebral malformation resulting from a defect in the early development of the prosencephalon. Its fetal prognosis is extremely poor, especially for the alobar form.

Objective: *Through the observation of a patient, we report the clinical, paraclinical and evolutionary aspects of this malformation.*

Observation: *In this case report, we report an alobaric holoprosencephaly diagnosed in the neonatal period and confirmed since birth.*

Conclusion: *Holoprosencephaly is a rare and fatal pathology with great etiological heterogeneity. In prenatal consultation, imaging (ETF, CT and MRI) allows an exhaustive lesion assessment of this pathology with an extremely reserved prognosis.*

Introduction

Holoprosencephaly is a rare cerebral malformation of multiple etiologies and often associated with evocative and specific facial anomalies (hypothelitism, cyclopia, ethimocephaly, cleft lip and palate). This pathology, resulting from a defect in the early development of the prosencephalon, has an extremely poor fetal prognosis, especially for the alobar form.

Objective

Through the observation of a patient, we report the clinical, paraclinical and evolutionary aspects of this malformation.

Observation

Female newborn born from a non-consanguineous marriage, hypothyroidism ATCD in the mother for 4 years continuously put under hormonal substitution during the first trimester by ignorance of the pregnancy, admitted on the first day of his birth to the pediatric emergencies CHU Med VI Marrakech for a cerebral malformation diagnosed in antenatal imaging highlighting a microcephaly with a large dilatation of a mono ventricular cavity, absence of the inter hemispheric scission and aging of corpus callosum. Clinical examination on admission showed facial dysmorphia with bilateral anophthalmia, cleft lip and palate, and externalization of brain tissue from the skull through a defect in the occipital bone. All four limbs were normal. The brain scan showed an alobar holoprosencephaly associated with an occipital meningoencephalocele. Chest and skeletal X-rays were normal, abdominal-renal and cardiac ultrasound showed no malformation. Karyotype revealed no genetic abnormalities. Surgery was not performed on our patient due to the complexity of the procedure. The newborn died on the 25th day of life.

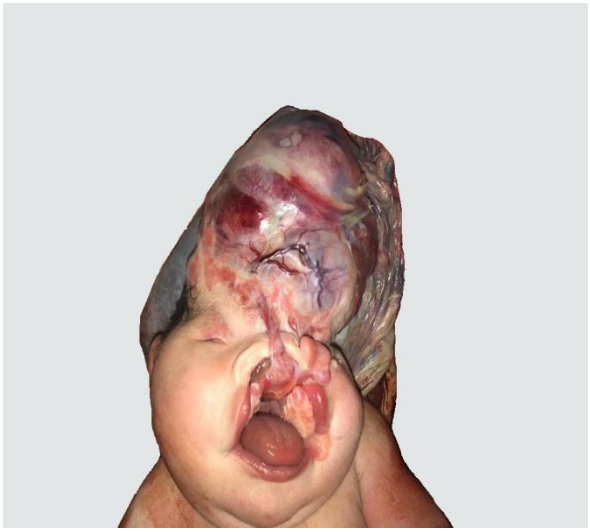

Figure 1: Front view

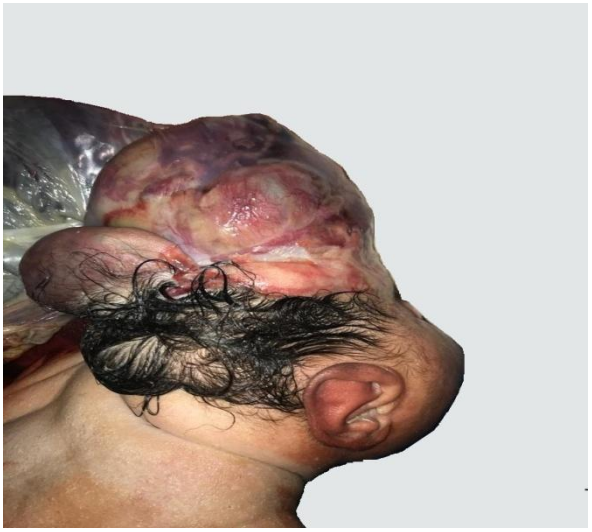

Figure 2: Side view

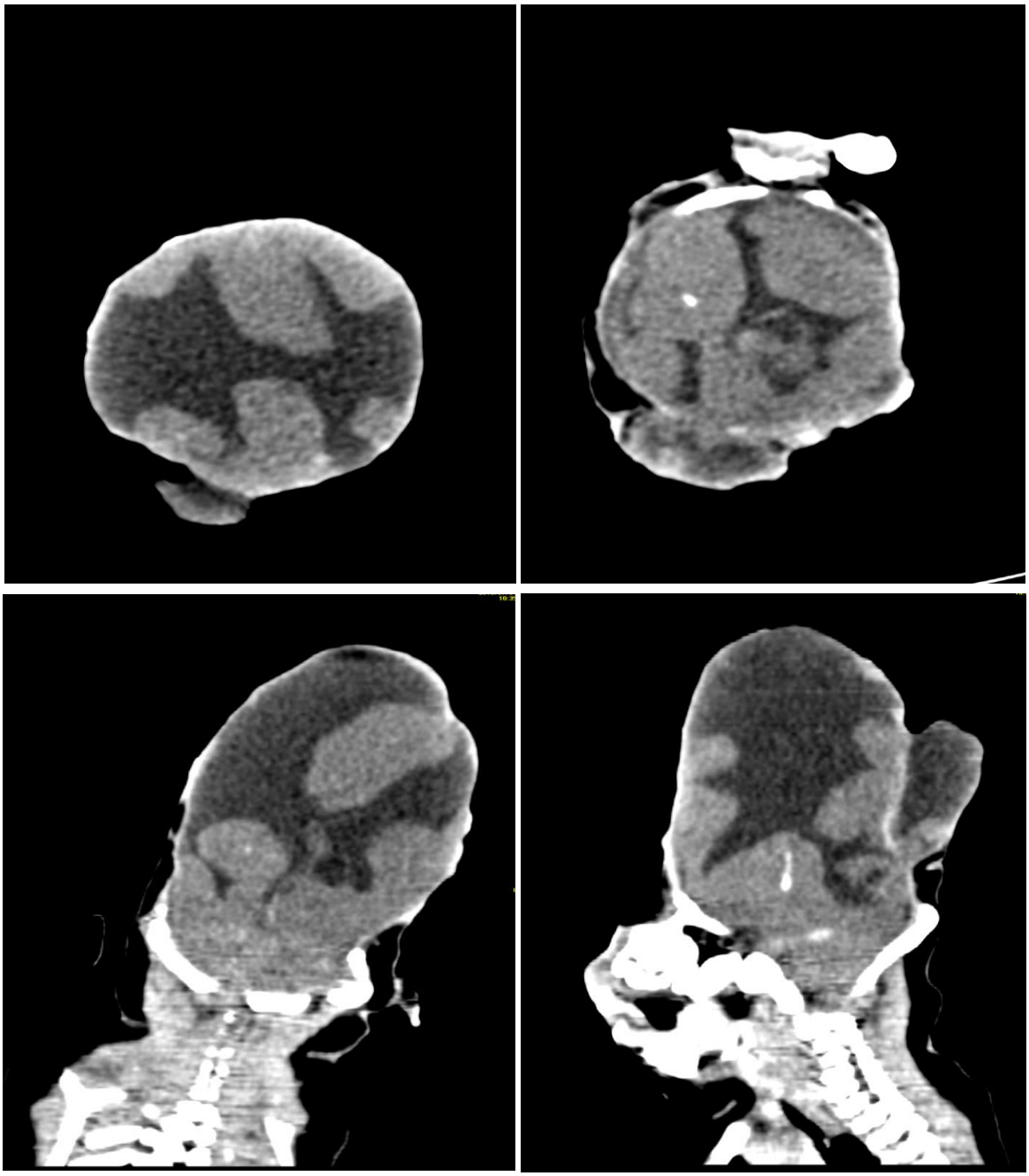

Figure 3: Brain scan: Alobair holoprosencephaly associated with an occipital meningoencephalocele.

Discussion

There are three classic anatomical forms of EH in order of increasing severity: lobar, semi-lobar and alobar EH. Another less severe form has been reported, the medial interhemispheric variant. The spectrum of HPE also extends to aprosencephaly/atelencephaly (at the extreme), schizoencephaly, and septopreotic HPE; the less severe forms known as microforms of HPE are characterized by midline abnormalities without the cerebral cleavage defect typical of HPE (see these terms). The spectrum of HPE covers a continuum of hemispheric separation defects with no clear distinction between the different forms with significant inter- and intra-familial variability. There is generally a correlation between the severity of facial and brain abnormalities. The main facial anomalies are, in decreasing degree of severity: cyclopia, proboscis, premaxillary age, moderate or bilateral cleft lip and/or palate, coloboma, retinal dysplasia, choanal atresia, pyriform sinus stenosis, hypolourism, and a single medial maxillary incisor; the face may be normal. Severe forms (associated with a particular chromosomal abnormality) are often fatal, and mortality correlates with the severity of brain malformations and other abnormalities. In surviving children, many associated manifestations are described: developmental delay, hydrocephalus, motor deficits, feeding and swallowing disorders, epilepsy, hypothalamic dysregulation. Endocrine disorders of pituitary origin such as diabetes insipidus are common.

Several authors agree on the association between maternal age and the occurrence of EPH [3, 4], and the absence of a causal relationship between gestational relatedness and the occurrence of EPH. The notion of consanguinity has been reported in the literature, as well as the incrimination of several genetic mutations [3,4].

The prevalence of EPH is less than 1 per 10,000 live births and a total prevalence of approximately 1.2 per 10,000 births [1, 2].

Holoprosencephaly results from a defect in the induction of neurectoderma by the prechordal plate during the third week of embryonic life, leading to an abnormality in the development of the prosencephalon consisting of a lack of evagination of the prosencephalic vesicles.

Alobair's HPE is the most severe form, in which the telencephalon consists of a holosphere containing a single ventricular cavity closed at its posterior part by a thin wall which gives it a pseudo-cystic appearance. The olfactory lobes are absent. The thalamis, small and rudimentary, are fused on the median line. Microcephaly is constantly present.

The prognosis was extremely poor. When a child with EHD is born for the first time, genetic counselling is of interest in order to assess the real risk of recurrence and to prevent its occurrence. Currently, molecular biology has made progress in identifying the genetic component of this brain malformation. Ultrasound screening remains a reliable means of pregnancy monitoring. In viable forms, surgery remains the only recourse. This is a complex surgery.

Conclusion

This clinical observation reflects a severe form of holoprosencephaly.

Bibliographic References

1. Dwight RC, Minal T, Jill AH. The etiopathologies of holoprosencephaly; Drug Discov Today Dis Mech; 2005.
2. Demeyer W, Zeman W, Palmer CG. The face predicts the brain: diagnostic significance of median facial anomalies for holoprosencephaly (arhinencephaly) pediatrics.
3. Fatnassi R, Turki E, Belhaj J, Labidi I, et al. Holoprosencephaly: pathogenesis, phenotypic characteristics - About four cases. Morphology. 2011.
4. Cuisset JM, Cuvellier JC, Vallée L, et al. Holoprosencephaly with neurogenic hypernatremia. Arch Pediatr. 1999 Jan;6(1):43-5. [Fetal malformation. EMC - Pediatrics. 2004 May;1(2):210-231.