Scholars Journal of Medical Case Reports
Abbreviated Key Title: Sch J Med Case Rep
ISSN 2347-9507 (Print) | ISSN 2347-6559 (Online)
Journal homepage: https://saspublishers.com

OPEN ACCESS

Radiology

Contribution of Fetal MRI in the Antenatal Diagnosis of Alobar Holoprosencephaly: A Case Report

Y. Bouktib[1*], K. El Hadri[1], D. Basraoui[1], H. Jalal[1]

[1]Radiology Department, Mother and Child Hospital Mohammed VI University Hospital, Faculty of Medicine and Pharmacy, Cadi Ayyad Marrakech University, Morocco

DOI: 10.36347/sjmcr.2023.v11i04.040 | **Received:** 25.02.2023 | **Accepted:** 12.04.2023 | **Published:** 18.04.2023

***Corresponding author:** Y. Bouktib
Radiology Department, Mother and Child Hospital Mohammed VI University Hospital, Faculty of Medicine and Pharmacy, Cadi Ayyad Marrakech University, Morocco

Abstract | **Case Report**

***Introduction*:** Holoprosencephaly is a rare cerebral malformation, of multiple etiologies and often associated with other malformations, particularly facial. It results from a defect in the early development of the prosencephalon. The fetal prognosis is extremely poor, especially in the alobar form. ***Objective*:** The aim of this work is to illustrate through this observation the contribution of fetal MRI in the antenatal diagnosis of this rare and serious pathology. ***Case Report*:** We report the case of a 26-year-old primigravida and primiparous parturient referred for suspected agenesis of the vermis and corpus callosum following a first ultrasound performed in the third trimester of pregnancy. The pregnancy was not followed up and the date of last menstrual period was unknown. A fetal MRI was performed showing: A mono-fetal pregnancy in cephalic presentation with fundial placenta, A single ventricle with posterior cystic expansion and laminated aspect of the cerebral parenchyma at the frontal level, A bilateral pyelo ureteral junction syndrome. ***Conclusion*:** Alobar holoprosencephaly is a rare and fatal pathology with a great etiological heterogeneity. It can be part of a polymalformative syndrome. It results from a cleavage anomaly of the prosencephalon into cerebral hemispheres. The antenatal diagnosis is based on obstetrical ultrasound completed by fetal MRI allowing an exhaustive lesion assessment of this pathology with an extremely reserved prognosis.

Keywords: polymalformative syndrome, prosencephalon, antenatal diagnosis, pregnancy, Alobar holoprosencephaly.

INTRODUCTION

Holoprosencephaly is a rare cerebral malformation, of multiple etiologies and often associated with other malformations, particularly facial. It results from a defect in the early development of the prosencephalon.

The fetal prognosis is extremely poor, especially in the alobar form.

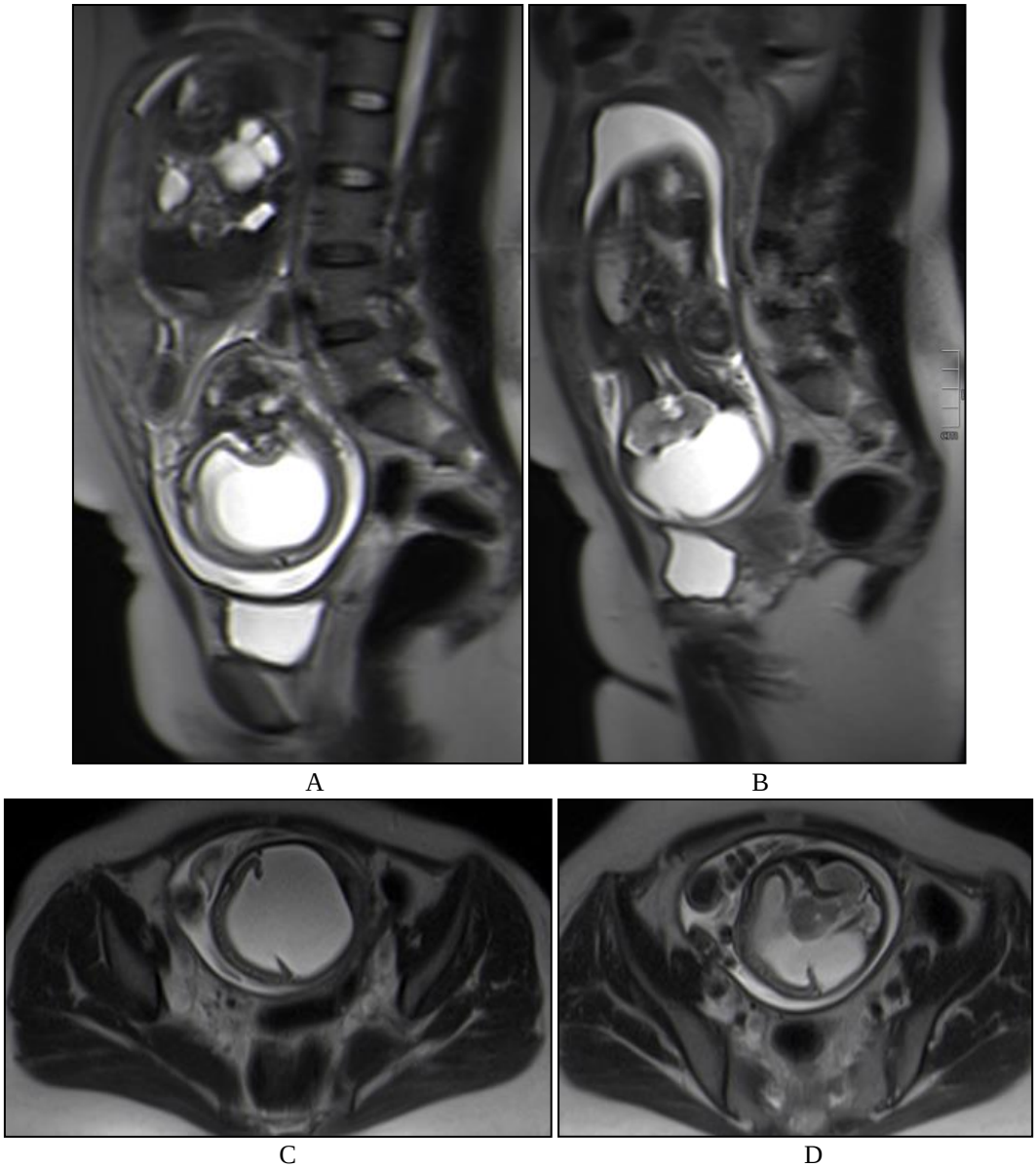
The aim of this work is to illustrate through this observation the contribution of fetal MRI in the antenatal diagnosis of this rare and serious pathology.

OBSERVATION

We report the case of a 26-year-old primigravida and primiparous parturient referred for suspected agenesis of the vermis and corpus callosum following a first ultrasound performed in the third trimester of pregnancy. The pregnancy was not followed up and the date of last menstrual period was unknown.

A fetal MRI was performed showing:

- A mono-fetal pregnancy in cephalic presentation with fundial placenta.
- A single ventricle with posterior cystic expansion and laminated aspect of the cerebral parenchyma at the frontal level.
- A bilateral pyelo ureteral junction syndrome.



Figures: Pregnancy of 34 SA. Alobar holoprosencephaly with bilateral pyeloureteral junction syndrome. A and B: sagittal sections, C and D: axial sections

- Single ventricle with posterior cystic expansion and laminated aspect of the brain parenchyma at the frontal level.
- Absence of medial structures.
- Posterior cerebral fossa with normal appearance.
- Bilateral pyelo ureteral junction syndrome predominantly on the left.

DISCUSSION

Holoprosencephaly (HPE) is a severe and complex congenital malformation of the brain associated with suggestive and peculiar facial anomalies [1, 2].

It is a rare disease with a prevalence of approximately 1.2 per 10,000 births [1, 2]. Its incidence is underestimated due to the abortive nature of the malformation and the minor forms that may go unnoticed. Several authors agree on the association between maternal age over 30 years and the occurrence of EH [3, 4].

The notion of consanguinity has been reported in the literature, as well as the incrimination of several genetic mutations [3, 4].

The holoprosencephaly results from a defect of induction of the neurectoderm by the prechordal plate, during the third week of embryonic life, which results in an abnormality of the development of the prosencephalon consisting of a lack of evagination of the responsible prosencephalic vesicles:

- Of the presence of a medial hemispheric mass replacing the two cerebral hemispheres.

- Of an absence of medial structures, including commissures.
- D an absence of differentiation or d an abnormal differentiation of structures derived from prosencephalic vesicles and diencephalic vesicle.

Based on the degree of individualization of the cerebral hemispheres, we distinguish three anatomical forms of EH: alobar, semi-lobar, and lobar [2]. The first two forms have a very poor prognosis, justifying early medical termination of the pregnancy [2, 4, 5]. Alobar HPE is 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 which gives it a pseudo-cystic appearance. The olfactory lobes are absent. The thalami, small and rudimentary, are fused on the midline. Microcephaly is consistently present.

D other types of CNS malformation may be associated with it such as atresia of the aqueduct of Sylvius, cerebellar hypoplasia, vermian agenesis, Dandy-Walker syndrome, and neural tube closure defects: encephalocele, spina bifida with myelomeningocele [2].

Ocular abnormalities are present in a quarter to a third of cases, single eye and synophthalmos, microphthalmia, cataract, retinal dysplasia, or iris or retinal coloboma may be observed [2].

The consistent association of facial anomalies testifies to the close embryologic relationships between the neurectoderm and the facial outlines. There are different phenotypes realizing a wide spectrum of severity. In a majority of cases, the severity of the facial anomalies reflects that of the cerebral malformations. Severe forms such as cyclopia and ethmorephaly usually correspond to alobar, sometimes semi-lobar, EH. Less severe forms are characterized by dysmorphic features [2, 6].

Median cleft lip and palate is one of the most frequent facial malformations in the context of holoprosencephaly (HPE) and requires a complete neuromorphologic and malformative workup [2, 6].

The antenatal diagnosis of EH is established by fetal ultrasound [5]. It is based on the association of intracranial signs and facial anomalies, especially in its complete form, but this association is not mandatory. In our patient, the antenatal diagnosis of the malformation was late, in the third trimester of gestation, due to a poorly monitored pregnancy, which contraindicated any therapeutic termination of pregnancy. Even if holoprosencephaly was diagnosed early, in Morocco therapeutic termination of pregnancy is still confronted with ethical, moral, legal and religious problems.

CONCLUSION

Alobar holoprosencephaly is a rare and fatal pathology with a great etiological heterogeneity. It can be part of a polymalformative syndrome. It results from a cleavage anomaly of the prosencephalon into cerebral hemispheres. The antenatal diagnosis is based on obstetrical ultrasound completed by fetal MRI allowing an exhaustive lesion assessment of this pathology with an extremely reserved prognosis.

BIBLIOGRAPHY

1. Dwight, R. C., Minal, T., & Jill, A. H. (2005). The etiopathologies of holoprosencephaly. *Drug Discov Today Dis Mech*, 529-537.
2. Demeyer, W., Zeman, W., & Palmer, C. G. (1964). The face predicts the brain: diagnostic significance of median facial anomalies for holoprosencephaly (arhinencephaly). *Pediatrics*, 34,256-63. PubMed | Google Scholar.
3. Fatnassi, R., Turki, E., Belhaj, J., Labidi, I., Regaya, B., Hidar, S., & Khairi, H. (2011). Holoprosencephaly: pathogenesis, phenotypic characteristics. About four cases. *Morphologie: Bulletin de L'association des Anatomistes*, *95*(310), 79-82. PubMed | Google Scholar
4. Cuisset, J. M., Cuvellier, J. C., Vallee, L., Ryckewäert, P., Soto-Ares, G., & Nuyts, J. P. (1999). Holoprosencephaly with neurogenic hypernatremia. *Archives de pediatrie*, *1*(6), 43-45. PubMed | Google Scholar
5. Moutard, M. L., & Fallet-Blanco, C. (2004). Fetal malformative neurological pathology. *EMC – Pediatrics*, 1(2):210-231. PubMed | Google Scholar
6. Mansouri Hattab, N., Lahmiti, S., Bouaichi, A., & Hiroual, A. (2011). Median cleft lip: one diagnosis masking another. *Arch Pediatr*, 18(2), 149-52. PubMed | Google Scholar