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Case Report

A Major Brain Malformation Revealed Late in the Third Trimester: Alobar Holoprosencephaly – A Case Report and Literature Review

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Abstract

Introduction: Holoprosencephaly (HPE) is a complex and rare brain malformation associated with facial abnormalities and has multiple etiologies. It results from a failure of medial cleavage of the prosencephalon in the right and left hemispheres. The prognosis is most often poor, particularly for the alobar form.

Case presentation: We report a case of alobar holoprosencephaly seen in 2024 in the Bechar region of Algeria. Our case was suspected based on an obstetric ultrasound at 35 weeks of gestation, which revealed a polymalformative syndrome. A brain MRI on the first day of neonatal life subsequently confirmed the diagnosis postnatally at 37 weeks of gestation.

Discussion: Holoprosencephaly affects the forebrain and face, causing neurological and facial manifestations. It is most often incompatible with life. Diagnosis relies on imaging, is usually delayed, especially in developing countries, and the prognosis remains poor.

Conclusion: Holoprosencephaly has an extremely poor fetal prognosis, particularly for the alobar form. It requires early diagnosis and multidisciplinary management, especially if termination of pregnancy is necessary, a situation that raises considerable speculation, particularly since this procedure is prohibited by law in Algeria.

Introduction

Holoprosencephaly (HPE) is a complex and rare brain malformation associated with facial anomalies (cleft lip and palate, cyclopia, hypotelorism, etc.). It results from a failure of medial cleavage of the prosencephalon into two cerebral hemispheres, occurring early during embryogenesis [1-2]. It is a rare fetal condition with diverse etiologies. It affects one in 15,000 to 16,000 births [3]. Three main anatomical forms have been described in the literature, with increasing severity: lobar, semilobar, and alobar. The alobar form is the most severe, characterized by a complete absence of separation of the cerebral hemispheres and frequently associated with major facial anomalies [1,4,5]. Early prenatal diagnosis of this malformation is essential to allow for appropriate and early obstetric management. We report the case of alobar holoprosencephaly suspected at 35 weeks of gestation and confirmed at 37 weeks of gestation by a brain MRI in a one-day-old newborn.

Case presentation

This is a 35-year-old patient, in her fourth pregnancy, with three uterine scars and no significant family or personal medical history. Her medical history revealed no exposure to any pathogen or toxin. No genetic testing was performed because this case was seen in a region far removed from highly specialized healthcare facilities, and the parents have a low socioeconomic status. The patient was referred to us for suspected threatened preterm labor associated with polyhydramnios and a fetal brain malformation discovered at

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35 weeks of gestation. Her medical history was unremarkable. On admission, the obstetric examination revealed an enlarged uterus with a fundal height of 36 cm. The fetal poles and fetal heart sounds were difficult to detect, and the cervix was closed on vaginal examination. Obstetric ultrasound suggested a diagnosis of holoprosencephaly, revealing a polymalformative syndrome consisting of significant polyhydramnios associated with hydrocephalus, suggestive of a giant monoventricle, absence of the midline, and fusion of thalamic structures along the midline (Figure 1). A cleft palate of the upper lip was also observed on 3D ultrasound (Figure 2). Maternal blood tests showed normal fasting and postprandial blood glucose levels. Serological tests for toxoplasmosis and rubella demonstrated residual immunity. A scheduled cesarean delivery occurred at 37 weeks of gestation, resulting in the birth of a live female newborn, with an Apgar score of 7/10 at one minute and 8/10 at five minutes, weighing 3600 g. After examination of the newborn in the operating room by the pediatrician, she was transferred to the neonatal unit for monitoring and further investigation. Neonatal examination revealed macrocephaly associated with a fetal malformation including a cleft lip and palate, and striking dysmorphic features. The head circumference was 41 cm with large fontanelles; the rest of the clinical examination was unremarkable, prompting the neonatologist to order a brain MRI on the first day of neonatal life, which revealed lobar holoprosencephaly characterized by a complete fusion of the median cerebral structures which were very thinned, a single giant median ventricular cavity, occupying the majority of the surface of the cranial cavity on a coronal section and the absence of the midline figure 3. On a sagittal section, the MRI had objected in addition to that of the coronal section: mass effect with compression of the residual cerebral parenchyma, a thinning of the cortical mantle and agenesis of the corpus callosum figure 4.

The first 24 hours of neonatal monitoring were marked by the occurrence of a seizure. Echocardiography with Doppler revealed no abnormalities other than minimal mitral regurgitation. Chest X-ray and abdominal ultrasound were unremarkable. On the fourth day of neonatal monitoring, she experienced seizures again, and treatment with phenobarbital (Gardenal) was initiated: an initial loading dose of 20 mg/kg/day, followed by a dose of 5 mg/kg/day after stabilization. A transfontanellar ultrasound showed triventricular hydrocephalus associated with craniosynostosis. On the 13th day of neonatal monitoring, physiological jaundice developed, requiring phototherapy. She was discharged on the 15th day of her neonatal life with authorization to breastfeed and a prescription for Depakine 200 mg syrup, the initial dose being 10 ml/kg/day in two divided doses for the first week, then 20 mg/kg/day for the second week, and finally 30 mg/kg/day for the third week. She was followed up in neonatology

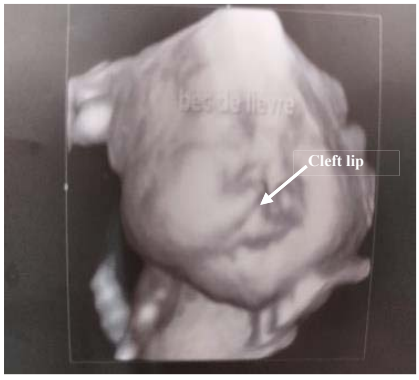


Figure 1: Obstetric ultrasound at 35 weeks of gestation: A cleft of the upper lip.

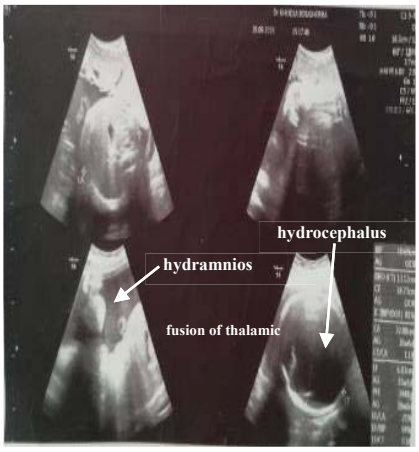


Figure 2: Obstetric ultrasound at 35 weeks of gestation: Multiple malformation syndrome associated with significant hydramnios.

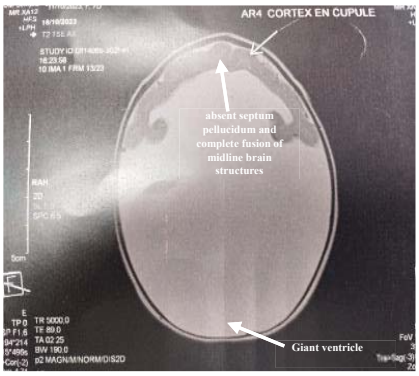


Figure 3: Coronal-section brain MRI showing complete fusion of the midline brain structures, which appear severely thinned; a single, giant, centrally located ventricular cavity occupying the majority of the cranial space on this coronal section; and the absence of a midline.

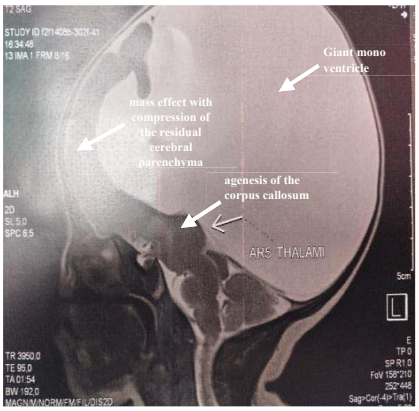


Figure 4: Sagittal brain MRI: a single, giant, midline ventricular cavity, mass effect with compression of the residual brain parenchyma, thinning of the cortical mantle, and agenesis of the corpus callosum.

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consultations while awaiting neurosurgical intervention, which did not occur. Later, an interview was conducted with the family, who stated that their child had died at four months of age from seizures at home, accompanied by hyperthermia of 41°C. We obtained explicit informed consent from the parents to publish the case and the radiological images, specifying that it was for scientific purposes.

Discussion

Alobar holoprosencephaly is a malformation affecting the forebrain. It is very rare but remains the most studied compared to other forebrain malformations [6,7]. Its prevalence is approximately 1 in 10,000 live births (1,2). Its pathogenesis is poorly understood and multifactorial, involving chromosomal, genetic, and environmental factors [6-9]. In our case, the etiology is undetermined because no genetic testing was performed. Several risk factors may be associated with alobar holoprosencephaly; these have been described in the literature, such as TORCH group transplacental infections, first-trimester bleeding, a history of miscarriage, and gestational diabetes [6]. However, none of these risk factors were found in our patient. Previous studies have shown that girls are more frequently affected, with a female-to-male ratio of 2:1, and this ratio increases with the severity of holoprosencephaly [5]. In our case, the newborn is indeed female; however, this does not allow us to generalize the result in any way, as it is only a single case.

The facial anomalies most frequently associated with holoprosencephaly are cyclopia, cleft lip, median or bilateral cleft palate, and hypotelorism [1,4,10,11]. The anomalies found in our newborn were a flattened nasal bridge, a cleft palate, and marked facial dysmorphism, suggesting a developmental defect in the cerebral and facial structures.

We suspected the diagnosis of holoprosencephaly thanks to the obstetric ultrasound which had shown a malformation syndrome at 35 weeks of amenorrhea and it was the brain MRI of the newborn which confirmed this diagnosis at 37 weeks of amenorrhea, our results agree with the literature regarding the means of diagnosis of this pathology [5,11,13,14].

The prognosis for alobar holoprosencephaly is poor, marked by an unfavorable course and high mortality [4,15]. In our case, tonic-clonic seizures began to generalize from the first day of neonatal life, with Depakine prescribed at a dose of 10 ml/kg/day in 2 divided doses for an undetermined duration.

During the newborn's monitoring, she presented with swallowing difficulties following oromotor dysfunction and cleft deformity; she required nasogastric tube feeding during the first three months of life and died in the fourth month of neonatal life in a picture of hyperthermia measured at 41°C, consistent with the very unfavorable evolution found in the literature.

A case was reported of a poorly monitored pregnancy where alobar holoprosencephaly was diagnosed late, and the newborn received inadequate care, partly due to the low intellectual level of both parents and the remoteness of specialized healthcare facilities. I even suggest that the pregnancy should have been terminated as soon as the diagnosis was suspected; unfortunately, Algerian law prohibits termination of pregnancy unless the mother's prognosis is at risk. I also propose creating multidisciplinary consultation meetings to discuss and decide on the legality of terminating pregnancy in cases of major fetal malformations incompatible with life, as is the case with alobar holoprosencephaly. This opinion in favor of terminating pregnancy in cases associated with alobar holoprosencephaly is consistent with the literature (2,8,14).

Conclusion

Alobar holoprosencephaly is a complex and rare fetal malformation with a high mortality rate, which must be detected and diagnosed early. It is legitimate to propose the creation of multidisciplinary consultation meetings to discuss and determine the legality of terminating pregnancy in similar cases.

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