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# Encephalocele Prenatal Diagnosis: About a Case

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**Abstract:- Encephalocele is the consequence of an incomplete closure of the cranial cavity. The prognosis depends on the topography and volume of the lesion. The antenatal diagnosis is made during the first trimester ultrasound in 80% of cases. We report two cases of encephalocele diagnosed antenatally by ultrasound in our department and managed immediately at birth by pediatric surgeons and neonatologists.**

## I. INTRODUCTION

Encephalocele is defined as the exteriorization of brain tissue and/or meninges outside the skull cavity through a congenital bony defect. The detection rate of cephaloceles in the first trimester has been estimated at 80% and it constitutes 5% of all severe structural abnormalities of the central nervous system (CNS)[1].

**Case 1:**

Patient aged 45 years, large multiparous, without notable pathological history, admitted in labor on a pregnancy estimated at 38SA + 5 days, the pregnancy was followed at the health center, without notion of morphological ultrasound. An ultrasound scan was performed, showing a severe hydrocephalus (figure 1), with a bi-parietal diameter of 118mm, with a brain parenchyma of 9mm, associated with a parietal encephalocele (figure 2), hence the indication for a caesarean delivery. Extraction of a male neonate, Abgard 10/10, birth weight 3300g, with presence of a macrocrania and an intact parietal encephalocele of 14cm (figure 3), neonate with good adaptation to the extrauterine life, neurological examination does not reveal any other abnormality, entrusted to the pediatric surgeon for an eventual cure of encephalocele.

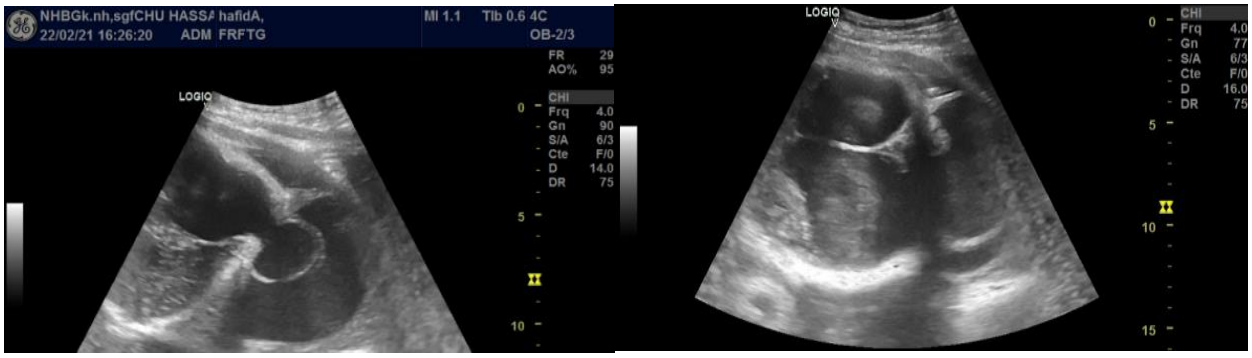


Figure 1, 2 : ultrasound images of a mass in the parietal bone, heterogeneous and predominantly anechogenic, evoking an encephalocele, and a severe hydrocephalus

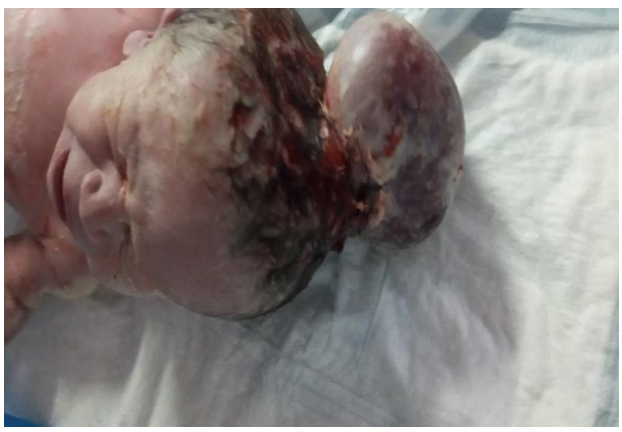

Figure 3 : aspect of the parietal encephalocele after delivery

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**Case 2:**

Patient aged 27 years, with no notable pathological history; second action; without notion of consanguinity, current pregnancy not followed (contracted in the period of pandemic Covid 19), admitted in active phase of labor on a pregnancy which is said to be at term, an obstetrical ultrasound was made before entering the delivery room and which objectified an evolutive mono-fetal pregnancy with presence of an anechne occipital mass in favor of an occipital encephalocele (figure 4) making 3cm, without other associated signs, notably no hydrocephalus or other neural tube closure anomalies, vaginal delivery was accepted, expulsion of a male newborn; the clinical examination confirmed the presence of an occipital encephalocele (figure 5) with a scalloped sac; with a normal neurological examination and good adaptation to extra uterine life. He was subsequently referred to the pediatric surgery department for management.

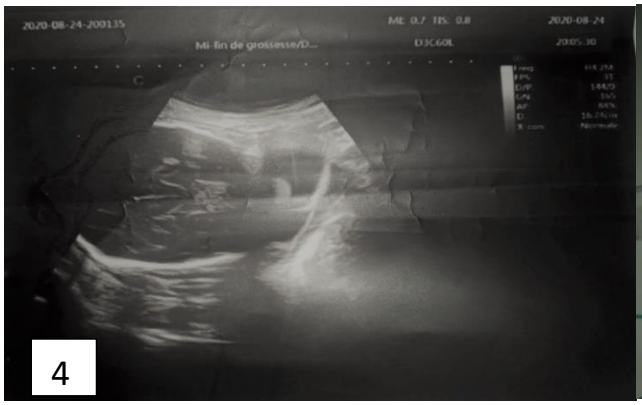



**Figure 4** : a heterogeneous occipital mass, with an anechogenic component, suggesting an occipital encephalocele.
**Figure 5** : aspect of the occipital encephalocele after delivery.

## II. DISCUSSION

Encephalocele affects 0.8 to 5 in 10,000 live births, [2] with marked geographic and ethnic variations. While the prevalence of neural tube defects has decreased significantly since the folic acid fortification that was recommended in the early 1990s, this question has been controversial with respect to the development of cephaloceles [3, 4].

For both patients folic acid supplementation was not taken nor were other supplements taken during their pregnancy.

with prognosis dependent on the amount of herniated brain tissue and other associated malformations. [5] More than 75% of encephalocele cases are occipital in location, while others may be frontal or parietal. [7] Prenatal diagnosis is usually made by ultrasound (US).

Schoner K, et all reported on a study done on neural tube defects; they found 17.7% of meningo-encephaloceles represent. [6] for our reported cases one is occipital of small size and was not associated with other malformation; and in the other case the encephalocele is of parietal localization which is a rare form of the encephalocele, and is associated with severe hydrocephalus and in any case the diagnosis is made by ultrasound.

Prenatal ultrasound to assess fetal morphology is usually performed early in pregnancy, between 11 and 14 weeks' gestation, and can detect a wide range of congenital anomalies, including neural tube defects. [7] Two-dimensional ultrasound can detect approximately 80% of encephaloceles, with diagnosis usually made in the second trimester. [5]

Although the onset of cephaloceles apparently occurs between 25 and 50 days of gestation (for lesions located anteriorly) and up to 60 days (for posterior defects), but the median GA of diagnosis is approximately 18 weeks of gestation according to the literature [8]**.**

for our two patients the diagnosis was late because they were not followed up; their pregnancies occurred at the same time as the pandemic period of COVID 19

The etiology of cranial hernias is not fully understood, but includes ethnic, genetic, and environmental factors. It is simply a failure of separation of the neural ectoderm from the surface ectoderm after closure of the rostral neuropore during the four weeks of gestation. [9].

Weichert J and all [10] have reported, 80% of defects are occipital versus 6% parietal. It is interesting to note that the latter are discussed as not constituting a genuine form of neural tube defect, but rather an origin of the defect stemming rather from environmental influences (e.g., reduced folic acid consumption) ; and It has recently been established that maternal passive smoking is an independent risk factor for three subtypes of NTDs (anencephaly, spina bifida and more strongly encephalocele) [11, 12].

In terms of management, a neurosurgical opinion is warranted. In a previous study published by Kiymaz et al, [13] factors such as lesion size, amount of neural tissue contained, ventriculomegaly, and other accompanying abnormalities negatively affected the prognosis of these

patients. The amount of neural tissue involved is associated with the severity of subsequent neurodevelopmental delays. It has also been reported that only about 17% of patients with encephaloceles had normal development, whereas 83% of patients had severe psychomotor developmental delays[14] Although the treatment is surgical correction, encephalocele remains a disorder with very high morbidity and mortality rates despite appropriate treatment.

## III. CONCLUSION

Encephalocele can appear isolated or as part of a polymalformative pattern. The antenatal diagnosis is mainly based on ultrasound. Advice to parents must be tailored individually and on a case-by-case basis; in a multidisciplinary approach with neuropediatricians, geneticists and neurosurgeons.

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