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Pre-Natal Diagnosis of Occipital Encephalocele: A Rare Case

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Abstract

Background: Encephalocele is defined as the externalization of brain tissue and/or meninges out of the skull through a congenital bone defect. It's a rare congenital malformation with a high risk of mortality and morbidity. Unfortunately, Prenatal diagnostic is still rare. Aims: The aim is to describe the main characteristic of this malformation and the optimal management to improve the prognosis. Case Presentation: we report the case of 32-year-old women diagnosed at 34 weeks gestation with a live fetus with occipital encephalocele. Conclusion: Through this case and the review of the literature we will highlight the diagnostic sources in order to improve the diagnosis and the management.

Keywords

Encephalocele, Prenatal Diagnosis, Tube Neural Malformation

1. Introduction

Encephalocele is defined as the externalization of brain tissue and/or meninges out of the skull through a congenital bone defect [1] [2]. It's a congenital malformation with a high risk of mortality and morbidity [3]. With an early prenatal diagnosis, the prognosis can be upgraded. Occipital encephalocele is the most common form of encephalocele. Through this case and the review of the literature we will highlight the diagnostic sources to improve the diagnosis and the management. We will also discuss the outcomes and procedure described in recent works. Proper diagnosis leads to proper treatment, important in the prognosis of this congenital malformation of brain.

2. Case Report

A 32-year-old, G2P2, 34-week pregnant woman without any familial or personal history, was consulted in our hospital for the second time during her pregnancy. She was from a low socioeconomic background with poor pregnancy follow-up. She had no complications during her pregnancy and didn't take any teratogenic substance. Her first child was born by vaginal delivery at term. Ultrasonography revealed a single live fetus with a breech presentation and an occipital encephalocele as we can see it in Figure 1. Moreover, the defect was about 18.9 mm (Figure 1).

We noticed the presence of cerebral structure in the cyst. No other abnormalities were found such as spina bifida. The neurosurgeon and the pediatric were noticed. A c-section was done at 39-week gestation allowing the birth of a newborn girl. The diagnosis was confirmed without any other abnormalities (Figure 2).

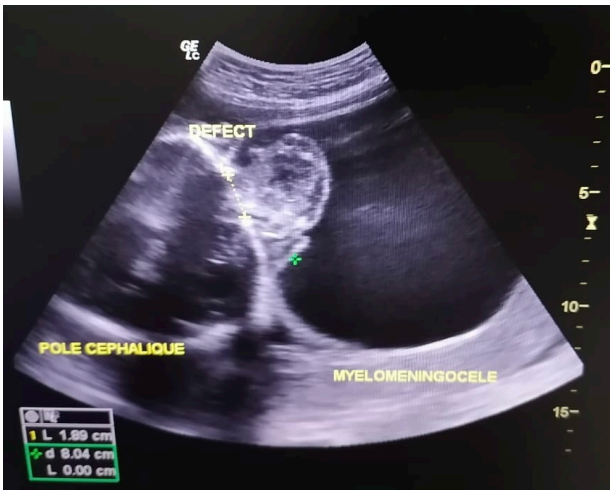

Figure 1. Ultrasound showing the encephalocele with the occipital defect.

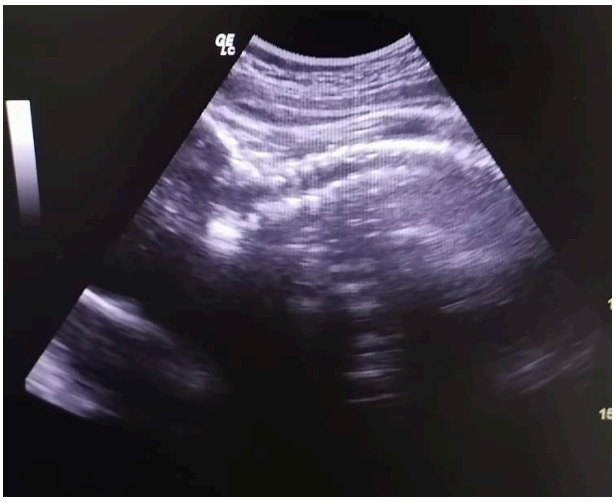

Figure 2. No other abnormalities were found in the ultrasound (like spina bifida).

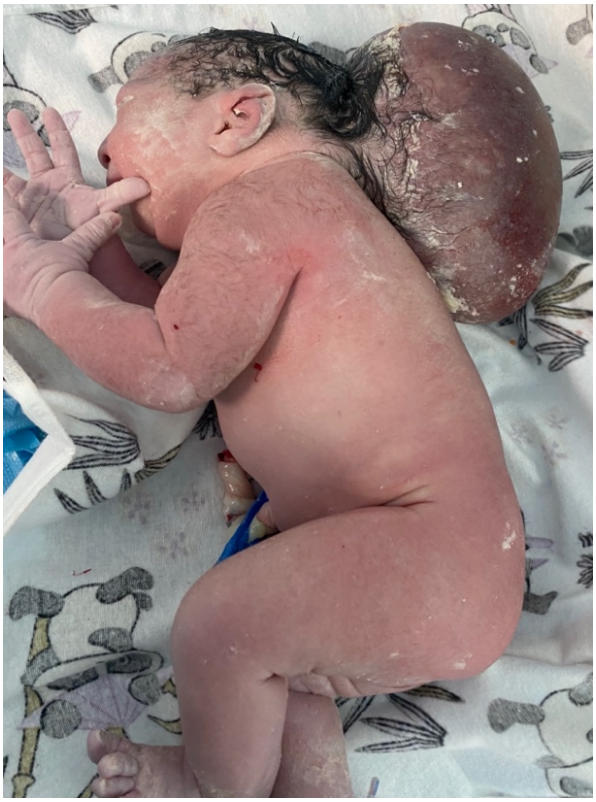

Figure 3. The newborn girl with an occipital encephalocele.

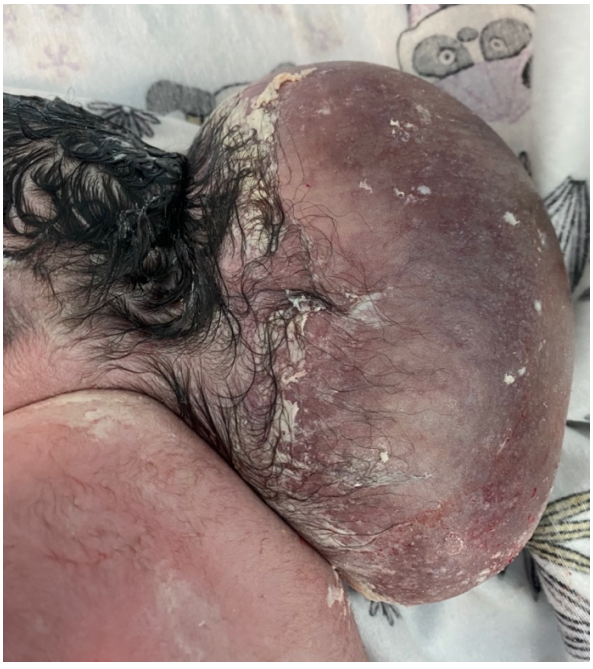

Figure 4. A picture showing the occipital defect.

Caesarean section was performed because of the breech position allowing the birth of a female newborn with an occipital encephalocele as suspected (**Figure 3** and **Figure 4**). A neurosurgery consultation was realized, and the operation was

done 3 months later. It consists in removing the bag and closing the defect. The fact that there was no brain tissue was easier. Post-operatively, the baby presented an infection of the wound without meningitis. One-year after her birth, she hasn't presented any notable complication.

3. Discussion

Encephalocele is defined as the externalization of brain tissue and/or meninges out of the skull through a congenital bone defect [1] [2]. It's a consequence of a failure to close the neural tube during the fetal development period [3] [4] [5]. This is one of the most severe neural tube abnormalities [1]. It is classified by location. It is divided into three major types: sincipital (frontoethmoidal), basal (trans-sphenoidal, sphenoethmoidal, transethmoidal, and spheno-orbital), and occipital [3].

The prevalence is estimated of 0.8 to 5 per 10,000 live births [1]. It represents about 10% - 20% of all craniospinal dysraphysmes craniospinal [1]. The mortality rate of encephalocele was 29%. The socio-economic level was poor in 70% of cases. The average maternal age was 27.8 years. All infants were born of large, poorly monitored babies. Prematurity was present in 45% of cases. The average size of the malformation was 4.75 cm and in 45% of cases, the size was greater than 10 cm [4] [5]. It's more frequent in females than in males [1], like in our case.

The etiology of encephaloceles is multifactorial including genetic and environmental factors. To date, the underlying mechanism behind congenital encephalocele is still uncertain, although it does imply a defective closure of the anterior neural tube. Some have suggested that the onset of the most severe lesions may occur before 26 days after conception, while less severe lesions primarily involving the skull or meninges may occur later [1]. Some teratogenic factors before 10 weeks' gestation may be involved [3].

According to literature review, encephalocele is usually diagnosed after birth sometimes with associated abnormalities like cleft palate, intellectual disability [3]. Prenatal diagnosis of encephalocele is carried out by maternal screening for serum a-foetoprotein levels and ultrasound [1]. The diagnosis is made easily and confidently from the ultrasound results during the second trimester and can also be made during the first trimester [2]. It's the most important way to make the diagnosis. Indeed, it detects about 80% of encephaloceles. The diagnosis is based on the detection of a cranial defect with a cerebral hernia of varying degrees. It appears as a cystic mass with a gyral pattern that is contiguous with the brain. Most cases occur in the occipital and rarely in frontal regions. The mass may be purely cystic, or it may contain echoes from brain tissue [1]. MRI during pregnancy is generally considered safe for the fetus, especially in the second or third trimester, thus facilitating to establish the prognosis of the diseases [1]. It can confirm the diagnosis suspected by ultrasound examination. But most importantly, its role is the evaluation of the remaining brain for other abnormalities. It can also contribute to post-delivery management schedule accurately by facili-

tating an opportunity for surgical repair before the condition leads to a serious complication [3]. MRI and ultrasound are then complementary [2].

Encephalocele can be part of genetic abnormality such as MECKEL-Gruber syndrome, which is associated with hexadactylia and bilateral nephrolomegaly. Other malformations can be associated such as hydrocephaly, corpus callosum agenesis, Arnold Chiari's malformation, microcephaly, craniostenosis [4], trisomy 18 or meningocele [1] [2]. 83% of patients with encephaloceles can have mental handicap and/or physical impairment. Seizure disorders are present in about 20% of infants with congenital encephalocele. Some complications can occur if the malformation is not diagnosed like intracranial hypertension, rupture, and superinfection [4]. The presence or absence of associated anomalies is the single greatest predictor of outcome.

Important questions arise after diagnosis [6]:

- Is this isolated or are there other associated anomalies.
- Does the patient wish to continue the pregnancy?
- Mode of birth and maternal/fetal implications of the choices.
- Postnatal management.

If encephalocele is voluminous, with severe microcephaly or other lethal abnormalities, termination of pregnancy may be indicated due to severe morbidity and mortality. Currently, the only effective treatment is restorative surgery [4]. Two techniques are possible: the conservative one and the no-conservative [4]. However, post-partum surgical treatment is appropriate for cases with a relatively small encephalocele and without other associated fetal abnormalities. The procedure consists essentially of removing the overlying bag and closing the defect, including the dural defect. Postoperative complications were mainly meningitis, surgical wound infection and hydrocephalus [4]. A third treatment option is prenatal surgery. This is well documented in several case series and reviews. It's an innovative method that consists in correcting the defect. Indeed, it may stop the progression of encephalocele. Furthermore, the outcomes seem better in this case [6].

Therefore, vaginal delivery may be considered if the lesion is relatively small. Another debatable indication is the potential bad outcomes (an amount of brain in the sac, other lethal abnormalities) of the disease. In this situation, it's better to avoid a c-section [2]. On the other hand, large fetal encephalocele may require a caesarean sectionin order to prevent a possible cephalopelvic disproportion [1].

The prognosis of neonates with encephalocele depends on the extent of the neural tissue hernia in the sac and the presence of associated abnormalities. Prognostic factors include bag size, neural tissue content, hydrocephalus, infection, and the presence of associated abnormalities. Lo and al reported that hydrocephalus and other intracranial abnormalities can predict the outcome of neurological development, but not the type of encephalocele [1].

4. Conclusion

The diagnosis of encephalocele should not be misunderstood because an early

diagnosis leads to correct management after birth. The main prognosis is the presence of brain tissues or other abnormalities. Intra-uterine surgery is a revolutionary treatment of the disease.

Consent

Written informed consent was obtained from the patient for publication of this case report and accompanying images. A copy of the written consent is available for review by the Editor-in-Chief of this journal on request.

Conflicts of Interest

The authors declare no conflicts of interest regarding the publication of this paper.

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