International Journal of Academic Health and Medical Research (IJAHMR)
ISSN: 2643-9824
Vol. 9 Issue 10 October - 2025, Pages: 35-37

Occipital Encephalocele: A Case Report And A Review Of The Literature

D.Rizki ,A.Benamar, N.Douzi, M.B Idrissi,N. Mamouni,S.Errarhay,C. Bouchikhi, A.Banani

Obstetrics and Gynecology Department, I, University hospital Hassan II Fez

***Abstract**: **Encephalocele** is a congenital malformation characterized by the herniation of brain tissue and/or meninges through a cranial bone defect. It may occur as an isolated anomaly or within a syndromic context, such as Meckel syndrome. Prenatal diagnosis is possible through ultrasound, which identifies a cranial defect associated with a herniated sac that may appear purely cystic or contain echogenic brain tissue. Management is surgical and involves resection of dysplastic neural tissue, coagulation of the choroid plexus within the malformation, and, when feasible, preservation of viable brain tissue without compression. Prognosis is primarily determined by the volume of herniated parenchyma and the presence of associated malformations. In this report, we emphasize the particular case of occipital encephalocele, focusing on its prenatal diagnosis and therapeutic management.*

Keywords: Occipital encephalocele, Antenatal imaging, pregnancy.

INTRODUCTION :

Encephalocele is a rare and severe neural tube defect, defined as the herniation of brain tissue and/or meninges through a congenital cranial defect. [1].

Occipital encephalocele, the most common form, results from abnormal closure of the cranial neural tube and may present as a meningocele, encephalocele, or meningoencephalocele.

Prenatal diagnosis is mainly achieved by ultrasound, where it appears as a cystic mass continuous with intracranial structures, and can be detected as early as the first trimester. Prognosis depends on the extent of neural tissue herniation and associated malformations, with frequently poor neonatal outcomes. [2].

We report a case of occipital encephalocele managed at HASSAN II Hospital in Fes, highlighting the contribution of antenatal imaging in diagnosis.

CLINICAL CASE :

This is a 17-year-old patient with no notable medical history, no known medication or toxic substance use, referred to our facility for a prenatal ultrasound to be evaluated for delivery planning. She came from a disadvantaged socioeconomic background and had received poor prenatal care. She had no complications during her pregnancy.

The obstetric ultrasound revealed a mass on the midline of the skull protruding in the occipital region making 110 * 150 mm

corresponding to an occipital encephalocele at 39 weeks of gestation (Figure 1).

A cesarean section was performed, to minimize the risk of sac rupture and delivery-related trauma, resulting in a female newborn with an Apgar score of 10/10 and a birth weight of 3000 g, presenting with an epidermal occipital encephalocele (Figure 2).

The infant appeared active. The neurological examination was normal. The rest of the external morphological examination was normal.

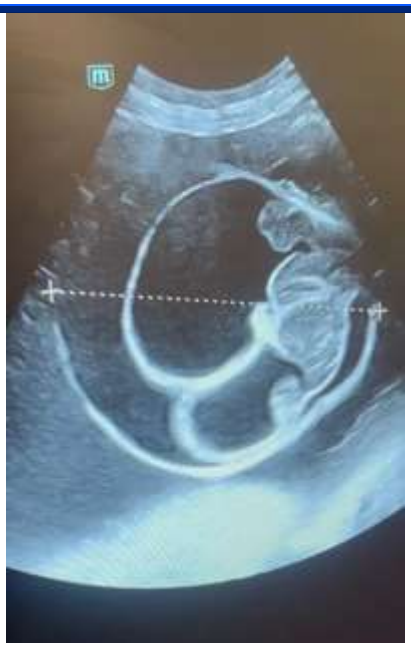

Figure 1: 39-week fetus, ultrasound shows a median occipital bone defect with cerebral parenchyma protrusion associated with isolated encephalocele.

Figure 2: Female newborn with Apgar score of 10/10 and birth weight of 3000 g presenting with a median occipital encephalocele.

DISCUSSION :

Occipital encephalocele is a rare congenital neural tube defect characterized by the herniation of brain tissue and/or meninges through a defect in the occipital bone. Compared to other neural tube defects, such as anencephaly or spina bifida, occipital encephaloceles are less frequent, representing approximately 10–20% of all craniospinal dysraphisms. Its etiology is heterogeneous and multifactorial, involving both genetic and environmental factors, although the precise pathophysiological mechanisms remain

incompletely understood. Severe lesions are believed to arise within the first 26 days post-conception, while milder forms, predominantly affecting the skull or meninges, may occur later in development [1,2].

renatal diagnosis relies primarily on ultrasonography, which allows visualization of a cranial defect with herniated brain tissue. Classic ultrasound features include a midline cranial mass, usually occipital, which can be cystic or contain echogenic brain tissue. Two-dimensional ultrasound detects most cases, therefore 3D ultrasound may facilitate early detection of fetal encephalocele and provide detailed visualization, thereby supporting prenatal diagnosis and genetic counselling. MRI is increasingly used to clarify the extent of the herniation and associated intracranial anomalies, particularly during the second or third trimester, and facilitates preoperative planning [3,4].

Occipital encephaloceles may occur in isolation or as part of a syndromic condition, such as Meckel-Gruber syndrome, which is classically associated with polydactyly and bilateral nephromegaly. Other associated anomalies, including trisomy 18 and holoprosencephaly, should be carefully assessed during prenatal imaging [5].

Postnatal management depends on the size of the encephalocele and the presence of associated lethal anomalies. Surgical treatment generally involves resection of the herniated sac, coagulation of dysplastic choroid plexus, and dural closure, with preservation of viable brain tissue when feasible. Prognosis is largely influenced by the amount of herniated neural tissue and the presence of other malformations. In cases with severe microcephaly or lethal anomalies, termination of pregnancy may be considered due to high morbidity and mortality [6].

In summary, advances in prenatal imaging, particularly ultrasound and MRI, have greatly improved early detection and characterization of occipital encephaloceles, allowing timely counseling and planning for postnatal surgical management, ultimately impacting clinical outcomes.

Delivery planning should be guided by the size of the encephalocele. Vaginal delivery may be feasible for relatively small lesions, while larger occipital encephaloceles typically require cesarean section to minimize the risk of birth trauma [1].

Several factors affect prognosis, including the sac size, the amount of herniated neural tissue, presence of hydrocephalus, risk of infection, and associated anomalies. Recent studies indicate that hydrocephalus and other intracranial malformations are important predictors of neurological outcome, whereas the encephalocele subtype alone does not significantly influence prognosis [7]. In these cohorts, cognitive impairment was observed in a substantial proportion of patients, with mild, moderate, or severe deficits reported in 11%, 16%, and 25% of cases, respectively [6]. Mortality remains significant, with a reported rate of approximately 29% for occipital encephaloceles, reflecting the potentially serious clinical course [1,6].

CONCLUSION :

Encephalocele can occur in isolation or as part of a polymalformatif syndrome. Prenatal diagnosis is based primarily on ultrasound, with other tests such as fetal MRI and AFP testing may be requested as a second-line investigations.

REFERENCES:

1. Markovic I, Bosnjakovic P, Milenkovic Z. Occipital Encephalocele: Cause, Incidence, Neuroimaging and Surgical Management. Curr Pediatr Rev. 2020;16(3):200-205.Parelkar SV, Joshi S, Gawde S, et al. Meckel-Gruber syndrome: a rare and lethal anomaly with occipital encephalocele, polycystic kidneys, and postaxial polydactyly. *J Pediatr Neurosci*. 2013;8(2):139-141.
2. Date I, Yagyu Y, Asari S, Ohmoto T. Long-term outcome in surgically treated encephalocele. Surg Neurol. 1993 Aug;40(2):125-30. doi: 10.1016/0090-3019(93)90122-h. PMID: 8362349.
3. Qudsieh S, Barbarawi MM, Qudsieh H, Barbaraw L. Giant encephalocele in newborns: prenatal diagnosis, management and outcome. Childs Nerv Syst. 2025 Jan 11;41(1):89. doi: 10.1007/s00381-025-06750-3. PMID: 39797973.
4. Liao, S., Tsai, P., Cheng, Y., Chang, C., Ko, H., & Chang, F. (2012). Prenatal diagnosis of fetal encephalocele using three-dimensional ultrasound. Journal of Medical Ultrasound, 20(3), 150–154.
5. Parelkar SV, Kapadnis SP, Sanghvi BV, Joshi PB, Mundada D, Oak SN. Meckel-Gruber syndrome: A rare and lethal anomaly with review of literature. J Pediatr Neurosci. 2013 May;8(2):154-7. doi: 10.4103/1817-1745.117855. PMID: 24082939; PMCID: PMC3783728.
6. Kiymaz N, Yilmaz N, Demir I, Keskin S. Prognostic factors in patients with occipital encephalocele. Pediatr Neurosurg. 2010;46(1):6-11. doi: 10.1159/000314051. Epub 2010 May 5. PMID: 20453557.
7. Da Silva SL, Jeelani Y, Dang H, Krieger MD, McComb JG. Risk factors for hydrocephalus and neurological deficit in children born with an encephalocele. J Neurosurg Pediatr. 2015 Apr;15(4):392-8. doi: 10.3171/2014.10.PEDS14192. Epub 2015 Feb 6. PMID: 25658248; PMCID: PMC8483274.