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Occipital Encephalocele in a 10-Day-Old Female Neonate: A Case Report and Review of Management

[1]Harshitha Bunga, [2]Kanchi Lavanya, [3]Sravya Sai Deepika Pandilla, [4]Bobbala Naga Naveen and [5]Marri Boyina Srikanth Yadav

[1,2]Department of Paediatrics, Osmania Medical College, 5-1-876, Turrebaz khan Rd, Troop Bazaar koti, Hyderabad, Telangana 500095, India
[3-5]Department of Paediatrics, Viswabharathi Medical College, PVVJ+Gfj, Kurnool Bellary Road, Penchikalapadu, Gudur, Andhrapradesh, 518467, India

ABSTRACT

Occipital encephalocele is a rare neural tube defect (NTD) characterized by the herniation of brain tissue through a skull defect, typically in the occipital region. This case report focuses on a 10-day-old female neonate presenting with occipital encephalocele, highlighting the importance of prenatal diagnosis, multidisciplinary management and preventive maternal care, particularly folic acid supplementation, in reducing the risk of NTDs. A 10-day-old female neonate (Twin 2) born to a mother from Bidar, Karnataka, presented with a sac-like protrusion from the occipital region of the skull, which was detected during a Targeted Imaging for Fetal Anomalies (TIFFA) scan at five months gestation. The mass progressively increased in size from the time of diagnosis until delivery. The infant was delivered via emergency lower segment cesarean section due to fetal bradycardia, weighed 2.1kg and had no immediate postnatal complications such as vomiting, seizures, or respiratory distress. A thorough physical and neurological examination revealed diminished reflexes and hypotonia. Investigations confirmed the diagnosis of occipital encephalocele, necessitating surgical intervention. The development of occipital encephalocele is associated with maternal risk factors such as folic acid deficiency, antiepileptic drug use and diabetes. Early prenatal detection via ultrasound or maternal serum markers, followed by a multidisciplinary approach to neonatal care, is essential for improving outcomes. Surgical closure of the defect and long-term monitoring are the mainstays of management. This case underscores the importance of early antenatal detection of neural tube defects and the preventive role of maternal folic acid supplementation. Multidisciplinary management and surgical intervention are crucial in optimizing outcomes for neonates with occipital encephalocele.

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Key Words
Occipital encephalocele, neural tube defects, folic acid

Corresponding Author
Kanchi Lavanya,
Department of Paediatrics, Osmania Medical College, 5-1-876, Turrebaz khan Rd, Troop Bazaar Koti, Hyderabad, Telangana 500095, India
Kanchilavanya3010@gmail.com

Author Designation
[1,5]Clinical Intern
[2,4]Medical Graduate

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INTRODUCTION

Neural tube defects (NTDs) represent a significant group of congenital malformations that arise due to improper closure of the neural tube during embryonic development, typically within the first 28 days of gestation. These defects can occur along any point of the neural tube, leading to malformations involving the brain, spine, or spinal cord. The incidence of NTDs is approximately 1.5 per 1,000 live births in India, with a wide spectrum of presentations ranging from anencephaly, spina bifida and encephalocele. Although the exact cause of NTDs remains multifactorial, maternal nutritional deficiencies, particularly folic acid and genetic predispositions, are known to contribute significantly to the risk of these malformations[1].

One subtype of NTDs is **encephalocele,** which occurs due to a defect in the bony structure of the skull, allowing the herniation of brain tissue and its coverings (meninges) through the defect. **Occipital encephalocele,** the most common form of this condition, specifically affects the posterior region of the skull, resulting in the protrusion of neural elements, usually covered by a thin membranous sac. The severity and extent of the encephalocele can vary widely, depending on the amount of brain tissue involved and the associated structural abnormalities of the brain[2].

Occipital encephaloceles are particularly significant as they pose immediate life-threatening risks due to the involvement of crucial brain structures, including the brainstem and cerebellum, which are vital for essential functions like breathing, heart rate and coordination. Additionally, the herniated brain tissue often undergoes ischemic damage due to mechanical disruption and insufficient blood supply, further complicating the prognosis[3].

Prenatal detection of occipital encephalocele has become increasingly possible with advancements in fetal imaging techniques such as the **Targeted Imaging for Fetal Anomalies (TIFFA)** scan, which allows for detailed visualization of fetal anatomy during pregnancy. This imaging modality is crucial for early diagnosis, providing the opportunity for parental counseling, planning for delivery and immediate neonatal intervention post-birth. Despite these advancements, the prognosis for large encephaloceles remains poor, especially when brainstem structures are involved[4].

This case report focuses on a 10-day-old female neonate with **occipital encephalocele,** born to a mother from a rural region with limited access to regular antenatal care. The case highlights the importance of early prenatal diagnosis, a thorough understanding of the clinical presentation and the role of prompt surgical intervention to improve survival and neurological outcomes. We will discuss the clinical features, diagnostic workup, differential diagnoses, and management strategies for this complex condition, as well as the role of preventive measures like maternal folic acid supplementation in reducing the risk of neural tube defects.

Case Presentation:

Patient Details: A 10-day-old female infant (Twin 2), born to Tabassum, a resident of Bidar, Karnataka, was referred to the Neonatal Intensive Care Unit (NICU) at Niloufer Hospital with a chief complaint of an opening defect in the occipital region of the skull, associated with a sac-like protrusion of neural tissue. The neonate was delivered by emergency lower segment cesarean section (LSCS) due to fetal bradycardia identified during a twin pregnancy.

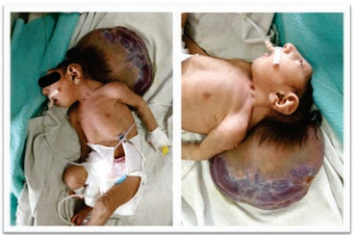
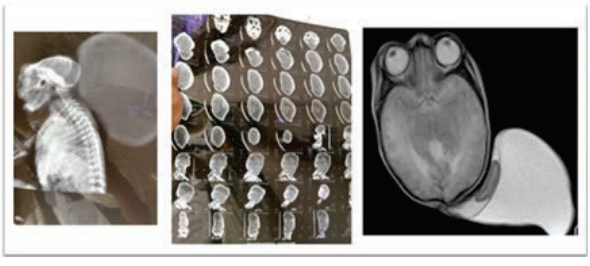


Fig. 1: 10-Days-Old Female Infant(Twin)

Complaints at Presentation: The primary presenting issue was the sac-like protrusion from the occipital skull defect, which had been identified during a Targeted Imaging for Fetal Anomalies (TIFFA) scan at five months of gestation. From the time of diagnosis to delivery, the size of the protruded mass continued to increase. Upon birth, the infant exhibited this neural mass protruding through the occipital defect.

History of Presenting Illness: Prenatally, the occipital encephalocele was diagnosed at five months gestation. The mother underwent emergency LSCS due to fetal bradycardia related to the twin pregnancy and the neonate cried immediately upon delivery.



Radiological Image 2:A Large Occipital Defect is Seen with Neural Tissue and Coverings Protruding Through it. Gyral Calcification is Noted in the Frontal Cortex. Additionally, there is Corpus Callosal Agenesis, Indicating the Absence or Malformation of the Structure Connecting the Two Cerebral Hemispheres.

The occipital defect with the protruding neural mass was noted at birth. The infant displayed no signs of vomiting, fever, seizures, respiratory distress, or other

concerning symptoms immediately after birth.

Birth and Perinatal History:
Antenatal History: The mother had irregular antenatal checkups, with no evidence of infections or complications such as Rh incompatibility or overt maternal diabetes. The mother did not consume alcohol or take antiepileptic medications such as Valproate or Carbamazepine during pregnancy.

Perinatal History: The infant was born via emergency LSCS at a birth weight of 2.1kg. The procedure was necessitated by fetal bradycardia. The baby cried immediately after birth, indicating good neonatal adaptation.

Feeding History: The infant was bottle-fed.

Immunization History: The newborn received routine vaccinations at birth, including BCG, OPV Zero and Hepatitis B.

Family History: There was no history of consanguinity, and the family had no known history of central nervous system (CNS) or neural developmental defects.

Socioeconomic History: The family belonged to the lower socioeconomic strata, classified according to the Kuppuswamy classification, which assesses income, education and occupation.

Physical Examination:
General: The infant was conscious, alert, moderately built and adequately nourished.

Vitals: Temperature: 36.5°C, Heart rate: 138/min, Respiratory rate: 56/min, Blood pressure: 86/52 mm Hg, Skin color: Pale to pinkish

Head-To-Toe Examination:
A sac-like protrusion of neural tissue through the occipital skull defect was noted.
No visible deformities were observed in the eyes, ears, nose, throat, chest, abdomen, spine, or extremities.

Cardiac Examination: Heart sounds S1 and S2 were present without any murmurs.

Respiratory Examination: Bilateral air entry was present and there was no deviation of the trachea.

Abdominal Examination: The abdomen was soft, with no palpable masses or defects.

Neurological Examination:
Mental Status: The infant was awake and conscious, but with neurological impairments associated with the encephalocele.

Cranial Reflexes:
Cranial Nerve II (Optic): The optic fundus appeared normal and the pupils were sluggishly reactive to light. Cranial Nerves III, IV, VI (Oculomotor, Trochlear, Abducens): Dolls' eyes were present, indicating functional brainstem reflexes.

Cranial Nerve V (Trigeminal): There was diminished rooting and sucking reflex.

Cranial Nerve VII (Facial): No facial asymmetry was observed.

Cranial Nerve VIII (Vestibulocochlear): A decreased response to sound was noted, suggesting possible auditory impairment.

Cranial Nerves IX, X (Glossopharyngeal, Vagus): Slight pooling of secretions was observed and the gag reflex was absent.

Motor System: The infant showed no spontaneous movements and limb tone was increased, with the limbs in an extended position. Hypotonia was noted in both the neck flexors and extensors.

Reflexes: Reflexes such as sucking, rooting, Moro and deep tendon reflexes were diminished, consistent with the neurological deficit associated with the encephalocele.

Investigations: Routine laboratory tests were performed, including:

Hemoglobin: 15.7 g/dL (within the normal range for a neonate).

White Blood Cell Count (WBC): 12,500 cells/cumm (indicating no acute infection).

Total Serum Bilirubin: 1.37 µmol/L (normal).

C-Reactive Protein (CRP): Positive (suggesting inflammation, though blood culture was negative for infection).

Blood Culture: Negative.

Coagulation Profile: Prothrombin Time (PT): 18.4 seconds, Activated Partial Thromboplastin Time (APTT): 35.6 seconds, International Normalized Ratio (INR): 1.54 (slightly elevated, indicating a mild coagulation disturbance).

Differential Diagnosis: Based on the clinical presentation, imaging and physical findings, the differential diagnosis included the following conditions:

- **Anencephaly:** Although considered, this was ruled out due to the presence of neural tissue within the encephalocele.
- **Meningoencephalocele:** Characterized by herniation of both meninges and brain tissue through a skull defect, this remained a possibility.
- **Hydrocephalus:** This condition involves the accumulation of cerebrospinal fluid within the brain's ventricles but was less likely without other supportive imaging findings.
- **Torticollis:** Unlikely, as this condition involves a shortening of the neck muscles, not a neural tube defect.
- **Occipital Encephalocele:** The final diagnosis, confirmed by clinical presentation and imaging, involved the herniation of brain tissue through an occipital skull defect.

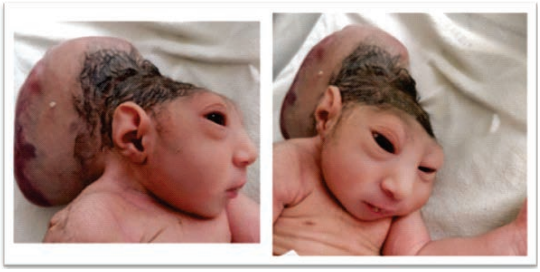


Fig. 3: Occipital Encephalocele

RESULTS AND DISCUSSIONS

Occipital encephalocele is a rare and severe form of neural tube defect (NTD) in which brain tissue herniates through a defect in the occipital region of the skull. This condition arises early in embryonic development, typically between the third and fourth week of gestation, when the neural tube fails to close properly. The skull defect allows for the protrusion of brain tissue, covered by meninges, creating a sac-like structure. In most cases, the herniated brain tissue becomes non-functional due to ischemic damage and mechanical disruption, leading to significant brain malformations. The extent of the encephalocele, along with the involvement of critical structures such as the brainstem, significantly influences the prognosis[5,6].

Several maternal and genetic risk factors are associated with the development of occipital encephalocele. **Maternal folic acid deficiency** is one of the most well-established risk factors for NTDs. Folic acid plays a crucial role in DNA synthesis, repair and cellular division during early pregnancy and its deficiency can result in improper closure of the neural tube. **Antiepileptic drugs,** particularly Valproate and Carbamazepine, have also been linked to an increased risk of neural tube defects. These medications interfere with folate metabolism, further predisposing the fetus to NTDs. **Maternal alcohol consumption** and **uncontrolled diabetes** are additional risk factors, as both conditions can interfere with normal embryonic development, increasing the likelihood of congenital malformations. Furthermore, **genetic mutations,** such as those in the MTHFR gene (methylene tetra hydrofolate reductase), can predispose individuals to folate metabolism deficiencies, exacerbating the risk of NTDs[7,8].

Risk Factors for Occipital Encephalocele:

- **Maternal folic acid deficiency**.
- **Use of antiepileptic drugs** (e.g., Valproate, Carbamazepine).

Maternal Alcohol Consumption:

- **Maternal diabetes.**
- **Genetic mutations** (e.g., MTHFR gene).

Early and accurate **prenatal diagnosis** is crucial for managing encephalocele. Diagnosis typically involves a combination of **ultrasound imaging**, which can detect the defect as early as 14-16 weeks of gestation and elevated maternal serum markers such as **alpha-fetoprotein (AFP)** and **acetylcholine esterase**. An elevated AFP level in maternal serum or amniotic fluid is a sensitive marker for neural tube defects, while acetylcholine esterase is more specific for open NTDs like encephalocele. Ultrasound allows detailed visualization of the skull defect and the herniated neural tissue, providing essential information for planning perinatal care and parental counseling[9].

The prognosis for occipital encephalocele varies widely depending on the size of the defect, the amount of herniated brain tissue and the involvement of critical structures such as the brainstem. **Large encephaloceles**, particularly those involving the brainstem, are often incompatible with life due to the critical role of the brainstem in regulating essential physiological functions such as breathing and heart rate. Conversely, **small encephaloceles** that do not involve vital brain structures may be amenable to surgical intervention, improving the chance of survival and functional outcomes[10,11].

The primary treatment for occipital encephalocele is **surgical intervention**. Surgery aims to remove the herniated, non-functional neural tissue and close the skull defect, ideally performed soon after birth. The success of the surgery depends on the extent of the encephalocele and the condition of the underlying brain tissue. In cases where brain tissue is not severely damaged and brainstem structures are intact, surgery can significantly improve the infant's chances of survival. However, even after successful surgery, long-term follow-up is essential, as children may experience neurological deficits, developmental delays, seizures, or hydrocephalus, requiring ongoing management by a multidisciplinary team[12,13].

Treatment Plan: In this case, the infant will require surgical intervention to remove the herniated neural tissue and close the occipital skull defect. The goal of surgery is to prevent further neurological damage and to promote healing of the skull defect. The infant's neurological status will need to be monitored closely postoperatively, with particular attention to motor and cognitive development. Long-term follow-up will be crucial in managing any residual neurological deficits and early intervention programs may be necessary to optimize the child's developmental potential[14].

CONCLUSION

This case of occipital encephalocele underscores the critical role of early antenatal detection in managing neural tube defects (NTDs). Prenatal screening tools, such as ultrasound and targeted imaging like TIFFA, play an essential role in identifying such congenital anomalies at an early stage, allowing for better planning and management. Early detection enables healthcare providers to counsel parents about the prognosis, potential interventions and necessary preparations for the birth and postnatal care of the affected infant. Moreover, this case demonstrates the importance of a **multidisciplinary approach** in the treatment of complex conditions like encephalocele. Surgeons, neonatologists, pediatric neurologists and other healthcare professionals must work together to manage the immediate and long-term challenges associated with the condition, including surgical repair, post-operative care and monitoring for neurological deficits.

Furthermore, the case emphasizes the **preventive role of maternal folic acid supplementation** in reducing the risk of NTDs. Maternal folic acid intake before conception and during early pregnancy has been shown to reduce the incidence of neural tube defects by up to 70%. This highlights the importance of public health initiatives and awareness programs that promote proper antenatal care, particularly for women in resource-limited settings where access to regular prenatal checkups and nutritional supplements may be limited. Routine supplementation of **0.4 mg of folic acid daily**, or higher doses in women with a history of NTD-affected pregnancies, is a simple yet highly effective preventive measure. Proper antenatal care, including folic acid supplementation, monitoring maternal health and ensuring timely interventions, is crucial to improving neonatal outcomes and reducing the burden of congenital anomalies like occipital encephalocele.

In summary, this case serves as a reminder of the necessity for comprehensive prenatal care, early detection and timely intervention to manage NTDs effectively. It also reinforces the importance of preventive strategies such as folic acid supplementation in reducing the incidence of these congenital conditions, highlighting the need for continuous public health efforts and improved access to maternal healthcare services.

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