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A Study of Congenital Cranial Encephalocele Cases in Egypt

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Abstract:

Background: Congenital cranial encephalocele, a neural tube defect characterized by the herniation of intracranial contents through a cranial vault defect, represents a significant public health challenge in developing nations. In these settings, its epidemiology is distinct, and patient outcomes are disproportionately poor. This review, augmented by original findings from a clinical cohort in Egypt, investigates the interplay between regional risk factors notably consanguinity, folic acid deficiency, and limited prenatal care and the resultant disease burden. It further evaluates the role of surgical intervention as a vital determinant of neurodevelopmental prognosis. Our analysis identified critical determinants of surgical outcome, including specific preoperative, intraoperative, and postoperative factors. The development of postoperative hydrocephalus was a pivotal complication. Notably, poor long-term outcomes delayed development or mortality in cases of our cohort were strongly predicted by the presence of concomitant cranial anomalies (e.g., Chiari malformation, corpus callosum agenesis, and Dandy-Walker malformation), large sac size, and significant intraoperative challenges. The evidence underscores the necessity of a dual strategy: aggressive public health initiatives for primary prevention must be coupled with the strategic strengthening of specialized neurosurgical and neurocritical care services. This integrated approach is essential to mitigate the substantial morbidity and mortality associated with this condition in resource-limited settings.

Keywords: Encephalocele; Neural Tube Defects; Epidemiology; Consanguinity.

Introduction:

Encephalocele is a congenital neural tube defect characterized by the herniation of intracranial contents including meninges, cerebrospinal fluid (CSF), and often brain tissue through a defect in the skull and dura mater. (Moore et al., 2018). The etiology of encephalocele is multifactorial, involving a combination of genetic predispositions and environmental influences. The global prevalence of encephalocele varies significantly by geographical region, ethnicity, and access to prenatal care and folic acid supplementation. The incidence is generally estimated at 0.8-4 per 10,000 live births worldwide. In developed countries, widespread folic acid fortification and prenatal screening have contributed to a reduction in neural tube defects, including encephalocele. However, in low- and middle-income countries where folic acid supplementation is less accessible, the incidence remains comparatively higher (Kumar et al., 2018). This study aims to clarify the unique epidemiological profile of encephalocele in the Egypt, analyze the modifiable risk factors that can underpin its high prevalence, and evaluate the critical role and outcomes of surgical management based on recent clinical evidence.

The diagnostic pathway for encephalocele, encompassing both prenatal and postnatal stages, is critical for prognosis and management planning. Prenatal screening primarily relies on fetal ultrasonography, a highly feasible method with reported sensitivity and specificity up to 94% and 98%, respectively, for detecting structural anomalies (Boyd et al., 2008). While three-dimensional sonography may offer superior lesion characterization (Cameron and Moran, 2009), prenatal MRI remains less accessible in resource-limited settings. Biochemical analysis of amniotic fluid, specifically acetylcholinesterase (AChE) levels, can confirm neural tube defects but is often reserved for complex cases in the era of advanced ultrasonography (Kooper et al., 2007). Postnatally, while the diagnosis is often clinically apparent, magnetic resonance imaging (MRI) is the modality of choice. MRI defines the anatomical extent of the lesion,

differentiates functional from gliotic neural tissue, assesses for concomitant hydrocephalus, and identifies other craniocervical anomalies. Furthermore, Magnetic Resonance Venography (MRV) is indispensable for delineating the relationship between the sac and major dural venous sinuses, thereby avoiding the risk of catastrophic intraoperative hemorrhage (Kotil et al., 2008).

The prevalence of encephalocele in Egypt is among the highest globally, a finding consistently reported in major epidemiological studies. According to a comprehensive systematic analysis by Blencowe et al., 2018 for the Eastern Mediterranean region, which includes Egypt, the prevalence of encephalocele is estimated at 3.7 per 10,000 births. A more specific, country-level analysis from the same research group calculated the prevalence in Egypt to be 2.09 per 10,000 births, a rate that is approximately seven times higher than that of the United States (Zaganjor et al., 2016). This elevated burden is driven by a combination of powerful and modifiable risk factors. Among these are consanguineous marriages, which significantly increase the probability of autosomal recessive genetic conditions, and maternal folate deficiency. These factors create a distinct risk profile that explains the significant difference in encephalocele prevalence between Egypt and many Western nations.

The predominance of preventable risk factors in the Egyptian cohort Shows an opportunity for primary prevention. The evidence is: periconceptional folic acid supplementation can prevent a significant proportion of NTDs (Berry et al., 1999). The finding that over half of the mothers in the study cohort did not receive folic acid is a direct call to action. A multi focused approach is necessary, including:

1. Public Health Education: Nationwide campaigns targeting women of childbearing age, especially in rural areas, on the importance of folic acid.
2. Food Fortification: Mandatory fortification of foods, such as wheat flour, with folic acid, a cost-effective strategy proven to reduce NTD incidence at the population level (Kancherla et al., 2022).
3. Genetic Counseling: Implementing community-based genetic counseling to educate populations with high consanguinity rates about the associated risks.

Addressing these modifiable factors is the most effective and sustainable strategy to reduce the burden of encephalocele and decrease the burden on surgical and rehabilitative services.
In established cases, timely and expert surgical intervention is the cornerstone of management, aiming to excise non-viable tissue, achieve watertight dural closure, and reconstruct the cranial vault to prevent infection and optimize neurological potential.

The technical principles, as detailed in the current study and the broader literature (Ozek et al., 2010; Jallo et al., 2018), involve careful preoperative planning with MRI/MRV, controlled resection of the sac, and meticulous layered closure.

The preoperative preparation mandates a complete neurological and general examination to identify associated congenital anomalies (Bui et al., 2007). The anesthetic management is particularly complex; patients, especially those with occipital encephaloceles and associated Chiari malformation, are prone to bradycardia and hemodynamic instability during laryngoscopy and positioning (Dey et al., 2007). A sudden intraoperative CSF leak can cause dramatic circulatory collapse, necessitating controlled drainage. Postoperatively, children face risks from central hypoventilation, hydrocephalus, and shunt malfunction, requiring monitoring in an intensive care setting to prevent fatal arrhythmias or respiratory arrest (Hamid & Newfield, 2001; Ganjoo & Kaushik, 1993). The absence of brain tissue in the sac remains the most favorable prognostic indicator, whereas the presence of gross, functional brain herniation portends a poorer outcome (Mahajan et al., 2011).

However, surgical outcomes are profoundly influenced by the very factors prevalent in low socioeconomic Countries. The studied cohort demonstrated that poor long-term outcomes (delayed development or mortality) were strongly predicted by:

- Disease Severity: The presence of other cranial anomalies (e.g., corpus callosum agenesis, Chiari malformation), huge sac size, and ulcerated skin were potent predictors of mortality and morbidity.
- Intraoperative Challenges: Findings of functioning brain tissue within the sac, hemodynamic instability, and significant blood loss were associated with dramatically worse outcomes.

- Postoperative Complications: The development of hydrocephalus, with its management being a major determinant of functional recovery.

This underscores that surgery, while life-saving, is not a adequate for better overall outcome. In resource-limited settings, the high prevalence of complex cases with associated anomalies means that surgical intervention often occurs within a clinical context already biased towards poorer prognoses.

Conclusion and Recommendations:

The challenge of encephalocele in developing countries like Egypt is a reflection of broader health system inequities. The condition's epidemiology is heavily shaped by preventable socioeconomic and nutritional factors, while its management is complicated by advanced disease presentation and high rates of postoperative complications.

A decisive reduction in the burden of encephalocele requires a shift from a purely surgical rescue model to an integrated approach:

1. Primary Prevention must be prioritized through mandatory folic acid fortification and comprehensive public health education.
2. Neurosurgical Capacity must be strengthened to manage complex cases and complications like hydrocephalus effectively.
3. Prenatal Diagnosis and Counseling should be expanded to allow for informed family decision-making.

Ultimately, tackling encephalocele demands a concerted effort that bridges public health policy and clinical excellence to break the cycle of disability and mortality associated with this congenital anomaly in the world's most vulnerable populations.

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