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CASE REPORT

Care management in the delivery room and neonatal intensive care unit: case report of a newborn with bilateral frontoparietal encephalocele

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Abstract

Nervous system defects affect 1 in every 4000 live births. Encephalocele, which consists of the herniation of the brain and/or meninges through a defect in the cranial vault, is among the main defects in the closure of the neural tube during embryonic development. The objective of this study was to report a case of ruptured bilateral frontoparietal encephalocele in a full-term child previously diagnosed since the 20th week of gestation. Born by vaginal birth, no resuscitation maneuvers were performed, no indication of greater investments due to malformations. 48 hours after birth, intensive care was initiated in the Neonatal Intensive Care Unit (ICU). During his stay in the ICU he received ventilatory support, phototherapy, surgical treatment for subpial transection of bilateral frontoparietal encephalocele, correction of cerebrospinal fluid fistula, cranial reconstruction for open acrania/dysraphism, anticonvulsant therapy, vasoactive drug support and antibiotic therapy. She was discharged to the ward after 40 days of hospitalization, without ventilatory support and on a full diet. Diagnosis is usually made via ultrasound in the second trimester of pregnancy. The prognosis is directly related to prenatal and postnatal care, the volume of herniated content, the brain tissue involved and whether there are other associated malformations. The indication of therapeutic interventions and the suspension of those already instituted often raise ethical dilemmas in neonatal care. Therefore, it is necessary to publish studies exploring this topic in detail.

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INTRODUCTION

Encephalocele is characterized by the protrusion of brain tissue beyond the normal limits of the skull through openings created by congenital bone defects or normal foramina or fissures in the skull, as more recently described[1]. The defect, typically found in the occipital region, can occur in isolation or association[2] with other congenital defects, contributing to various syndromes (20.5%)[2,3]. This condition may be linked to complications such as microcephaly, hydrocephalus, and Chiari malformation type III, among others (including facial, renal, and cardiovascular issues).[2]

Encephalocele is often classified as a neural tube defect (NTD), with anencephaly and spina bifida being the most common types in this category. Anencephaly, marked by the absence of the cranial vault and cerebral hemispheres, is a severe and fatal anomaly that occurs during the first weeks of embryogenesis. In contrast, spina bifida presents as a defect along the vertebra's midline, exposing the spinal canal's elements, and originates in the third or fourth week of embryonic life.[4]

Recent studies suggest that encephalocele may involve different basic mechanisms than spina bifida and anencephaly, related to specific phases of central nervous system embryology. Neural tube closure can occur through two models during embryogenesis: bidirectional closure (known as "the zipper model") and closure at multiple sites, which accounts for the variation in the anatomical location of encephalocele.

Additionally, genetic studies have shown discrepancies in concordance among siblings and in the prevalence of congenital anomalies between spina bifida, anencephaly, and encephalocele.[5] The risk of recurrence is greater in cases of spina bifida, highlighting the presence of varying risk factors among these conditions.[6]

The following known risk factors stand out: previous generations with neural tube defects, family history, teratogenic agents, maternal hyperthermia, gestational diabetes, maternal age over 40 or under 19 years, the use of certain medications during pregnancy (such as valproic acid and carbamazepine), smoking, alcohol use, and radiation exposure. Maternal folic acid deficiency also plays a significant role.[7]

The prevalence of encephalocele varies by region, typically occurring in about 1 in 1,000 live births. Ultrasonography (USG) is the preferred method for prenatal diagnosis, while complementary techniques such as skull X-ray, computed tomography, and magnetic resonance imaging may be necessary. The anatomical location of the encephalocele, along with other factors, affects treatment and prognosis, emphasizing the importance of prenatal diagnosis in determining the clinical approach.[8]

CASE DESCRIPTION

A female baby was born on March 3, 2021, with an APGAR score of 4/4/8 after 40 weeks and two days of gestation. She weighed 2,265 grams, measured 41 cm in length, had a head circumference of 26 cm, and blood type A-. During prenatal care, obstetric ultrasounds conducted around the 20th week indicated signs of a central nervous system malformation, which was confirmed by subsequent imaging tests shown in Figure 1 (authorized by the family). At delivery, the baby presented with a large, ruptured parietal encephalocele. Approximately 48 hours after delivery, while still in the obstetric center, the baby experienced difficulty breathing, apnea, and seizures, necessitating ventilatory support and transfer to a neonatal intensive care unit (NICU).



Figure 1. Obstetric ultrasound showing a fetus without a formed skull cap and brain parenchyma. Source: Author's archives, 2021.

The mother, a 19-year-old with blood type A+ in her first pregnancy, received an early gestational diagnosis after about ten prenatal consultations. She reported no use of medication, alcohol, tobacco, or other substances. The father has hypertension. An obstetric ultrasound performed at 36 weeks and four days of gestation did not provide an accurate characterization of the cephalic segment or differentiate between anencephaly/acrania and severe microcephaly.

On physical examination upon admission to the NICU, the newborn presented in poor general condition, exhibiting mild dyspnea in room air, appearing pale, hydrated, and jaundiced (Kramer's zone IV). The findings included symmetrical vesicular breath sounds, absent adventitious sounds, and rhythmic, normal heart sounds without bruits. The abdomen was assessed as normal with normal tone. The newborn was active and responsive to stimuli. She had a

large encephalocele with a fistula, draining a small amount of purulent secretion (Figure 2). A complete blood count did not indicate infection. A contrast-enhanced computed tomography scan of the head revealed a high parietal encephalocele with exposure of parenchymal tissue, meninges, and vascularity, as illustrated in Figure 2 below.



Figure 2. Newborn with a large encephalocele with a fistula and related computed tomography scan. Source: Author's archives, 2021.

Serology tests conducted on the mother were negative. Her blood count indicated mild microcytic, hypochromic anemia, with normal ferritin, folic acid, and vitamin B12 levels. The newborn's treatment included ventilatory support, phototherapy, anticonvulsant therapy, antibiotics, transfusion, and surgery. The neurosurgical intervention was carried out in two stages: the first involved a craniectomy and drainage of the cyst, followed by a subpial transection of the bilateral frontoparietal encephalocele, correction of the cerebrospinal fluid fistula, and cranial repair for the acrania/open dysraphism. The procedure was executed using a multidisciplinary approach with a specialized medical team. Despite the risks, complications were either avoided or managed, enabling the newborn to be discharged from the Intensive Care Unit 42 days after birth in good condition, allowing continued care in a ward bed.

DISCUSSION

When analyzing maternal risk factors, considering the specific social, political, and economic characteristics of developing countries, such as Brazil, is essential. In this context, potential teratogenic risks are associated with factors such as low educational attainment and income levels, high incidence of infectious diseases, limited resources for health and research, late gestational diagnosis, uncontrolled use of medications and narcotics, and the legal prohibition of terminating pregnancies. In the case presented, many of these risks are evident.[9]

Folic acid deficiency is recognized as a risk factor for neural tube defects, yet its specific relationship with subgroups like anencephaly, spina bifida, and encephalocele remains under investigation. While the exact role of folic acid in neural tube development is not fully understood, it is known that this complex B vitamin plays a crucial role in metabolic reactions necessary for synthesizing nucleic acids. However, recent studies show that in specific populations, the prevalence of encephalocele did not decline significantly despite increased maternal folic acid intake, which contrasts with the trends observed for anencephaly and spina bifida.[10]

Although the maternal serum folic acid levels were within the expected range, they did not contribute as an additional risk factor. Folic acid supplementation is considered essential, especially given the low adherence to medication in Brazil. The mandatory fortification of wheat and corn flours with iron and folic acid, implemented in 2004, aims to reduce the prevalence of maternal anemia and neural tube defects.[11]

It is important to note that excessive consumption of synthetic folic acid may be linked to vitamin B12 deficiency, which increases the risk of complications such as repeated miscarriages and multiple births. Preconception folic acid supplementation is recommended, with varying dosages for women with and without a history of neural tube defects.[12]

The surgical treatment of the encephalocele involved a multidisciplinary approach with a specialized medical team; despite the risks, complications were either avoided or treated, allowing the newborn to be discharged from the ICU in good condition and to continue receiving care in a ward bed.[13]

Throughout the delivery of care, the medical team, recognizing the complexity of the case, proposed palliative care to ensure comfort and quality of life. However, the mother was resistant to this suggestion, caught in a whirlwind of emotions that included unconditional love for her baby and the struggle to accept such a fragile reality. With its physical and emotional consequences, encephalocele presents a challenge that has exceeded medical understanding.[13]

Although neural tube defects have a varied etiology, with several mechanisms associated with their origin, most cases are linked to the interaction of multiple genes and environmental factors, characterizing a multifactorial inheritance. Prenatal diagnosis is significant in assessing the prognosis and providing appropriate genetic counseling.

The material and social costs associated with the complications arising from the lesions caused by the neural tube defect led to the review of potential factors that could improve or complicate the patient's clinical symptoms. Simple preventive measures can lead to complications, increased costs, and permanent sequelae if overlooked. In the neonatal intensive care unit, the mission extended beyond ensuring the patient's right to comprehensive and appropriate treatment; it also included providing psychosocial support for the family and guidance on implementing palliative care. In attempts to discuss palliative care with the mother, the team recognized

her resistance as a reflection of the instinctive nature of maternal love.

The medical team, attuned to the mother's distress, aimed to foster open and empathetic communication. They explored her expectations and wishes while providing clear information about her daughter's condition. This ongoing dialogue created a bridge of understanding between the medical team and the mother, consistently prioritizing the child's best interests.

It is essential to recognize that the mother's resistance to palliative care is not just a barrier to overcome but an expression of deep love and attachment. The medical team's role goes beyond clinical treatment; it involves emotional support, respect for family decisions, and searching for solutions that provide dignity and comfort.

In this context, the challenge was finding a delicate balance between offering the best care and respecting the family's choices. Compassion and empathy became essential tools in creating a family-centered care environment where the mother, child, and medical team could collaborate to provide the highest possible quality of life, regardless of the difficult choices that had to be made.

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