A Rare Association of Right-sided Congenital Diaphragmatic Hernia and Encephalocele: A Case Report

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ABSTRACT

Background: This is a case report regarding a 2051-gram female newborn affected by right-sided congenital diaphragmatic hernia (CDH) presenting with encephalocele in the occipital region.

Case report: The newborn was delivered by a 38-year-old mother from Darmian city, a rural district located in South Khorasan province, Iran.

Conclusion: The CDH is an abnormality that rarely occurs with an approximate ratio of one in 3000 live births. It has been reported that 85% of the infants affected by this condition are left-sided and the classic posterolateral or Bochdalek hernia is the most common form. An incidence of 40%–50% has been reported regarding the other malformations associated with CDH, the most common of which are those involving the central nervous system. Some studies reported other rare associated abnormalities, including hepatopulmonary fusion, hypoplastic left heart syndrome, left heart hypoplasia, duodenal atresia, malrotation, and anorectal malformation. This case report aimed to mention encephalocele as another complication, which has not been reported for the CDH.

Keywords: Congenital, Diaphragmatic, Encephalocele, Hernia, Newborn

Introduction

Congenital diaphragmatic hernia (CDH) is an abnormality found in one per 2500 newborns with a survival rate of 67% (1). Right-sided, left-sided, and bilateral CDH occur in 10%, 85%, and < 5% of the cases with this problem (2, 3). Approximately 50%-60% of the affected individuals have isolated CDH and the remaining have the complex form occurring with additional malformations or as part of a single gene disorder and/or chromosome abnormality (1, 4).

One of the primary characterizations of the CDH is failure in proper diaphragm formation during the course of embryogenesis (5). This incomplete diaphragm formation allows abdominal contents to herniate into chest leading to a mass-like effect that impedes lung development. Clinical presentation of this case ranges from asymptomatic to serious respiratory or gastrointestinal symptoms and occasional hemodynamic instability (5).

The broad spectrum of severity in patients with CDH depends on the degree of pulmonary hypoplasia and hypertension. Posterolateral or Bochdalek hernia (BH) is known as the most common type of hernia (>80%) with the majority occurring on the left side (85%) and less frequently on the right side (13%) or bilateral (2%) (6).

Encephaloceles are characterized by herniation of intracranial structures through a partial deficiency in the skull. The encephaloceles are a rare incidence (i.e., 0.01%-0.02% in all live births) subclassified into atretic cephalocele, meningocele, meningoencephalocele, and meningoceles according to the contents of herniation (7, 8).

Prenatal diagnosis of the association between a right-sided CDH and encephalocele has not been reported in the medical literature as searched in the Medline database.

Case report

A 38-year-old female from Darmian, South Khorasan province of Iran with a history of seven...
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gravidities and seven parities (three live and three dead) delivered a 2051-gram female newborn with one- and five-minute Apgar scores of 3 and 6. The 38-year-old woman had no abnormal medical history and was on antenatal iron supplement. The parents had no familial relationship and the routine antenatal blood tests of the mother, including hepatitis B, rubella, and TORCH screen were all reported as negative.

Due to their low socioeconomic status, only one normal ultrasonography was conducted on 13 weeks of gestational age. After birth, the infant had an obvious 2×2 cm encephalocele in the occipital region with all the lesion being covered by skin. As the result of encephalocele, cyanosis, and hypothermia, the infant was intubated and was transferred from the operating room to the Neonatal Intensive Care Unit (NICU) immediately.

The first biochemistry tests of the infant showed pH, PaCO₂, PaO₂, and HCO₃ as 7.02, 55.8%, 50.7%, and 14.4, respectively. Moreover, the hematologic profile entailed white blood cell, neutrophil percentage, lymphocyte percentage, hemoglobin, platelet count, and blood group of 16.3 1000/mm³, 41.8%, 50.4%, 17.2 g/dl, 183000/mm³, and O⁺, respectively. A portable chest X-ray revealed a fairly large diaphragmatic hernia with multiple loops of bowel in the right side of the thorax (Figure 1).

During the NICU hospitalization period, the infant did not respond to mechanical ventilation due to the pulmonary hypoplasia. Despite the increase in the ventilation setting, the results were not favorable and condition of the patient deteriorated leading to death within an hour after the birth.

Given the fact that during hospitalization the condition was unstable and brain magnetic resonance imaging was not performed during mechanical ventilation in the designated unit, brain condition of the infant was not clearly understood. In addition, unstable hemodynamic status of the newborn did not allow surgeons to recover the encephalocele lesions. Unfortunately, the parents did not agree with neither taking photos from the neonate nor performing autopsy in order to find the incidental internal visceral abnormalities.

Discussion

In this report, a very first medical case of right-sided CDH and encephalocele was presented. There had not been any reports found with this content in the English literature. Nevertheless, Ali and Ahmed were able to find 18 cases of encephalocele and myelomeningocele associations since 1975 until their own study was published (2).

CDH is a lethal birth defect with a recently reported prevalence of 0.17-0.57 per 1000 live births. A meta-analysis of 12 studies compiled by Skari et al. (9) stated the pooled frequency of right-sided CDH to be 8%. The hidden mortality (deaths before admittance to a treatment center) has been shown to influence mortality rates in CDH. However, its validity has been questioned recently (10-13).

Presence of the associated major malformations has been reported as a negative prognostic factor although the intensity of this effect on mortality rate has not been settled (14, 15). An augmentation in the mortality rate in the right-sided, compared to the left-sided CDH has been reported (11, 16, 17). However, other studies have found no such influence (10, 18) or an opposite effect (19) on the mortality rate.

The neonates reported by Akinkuotu et al. had a significant clinical problem occurring rarely as one case per 2500-3000 human births (20). This condition continues to have a high mortality rate due to the lethal combination of pulmonary hypoplasia and pulmonary hypertension (21, 22).

Presence of the associated major malformations is considered as a negative prognostic factor worldwide. Furthermore, an increased mortality rate has been indicated in the right-sided versus
left-sided CDH (23). Associated chromosomal abnormalities have a reported prevalence range of 9.5%-21% (9, 24). Occurrence of duodenal atresia, malrotation, and anorectal malformation has been previously reported in two patients (25). Moreover, right-sided CDH (26) and gallbladder agenesis have been reported along with duodenal atresia (27). Chen et al. (28) reported a rare case of a neonate with simultaneous VACTERL association and right-sided CDH.

Hepatopulmonary fusion is an uncommon congenital malformation associated only with a right-sided CDH (29). We found some rare associated anomalies, such as hepatopulmonary fusion and hypoplastic left ventricular syndrome in the literatures (29-31). Therefore, we recommend that encephalocele could also be regarded as a rare association to the list of abnormalities.

Conclusion
It seems there is an association between congenital diaphragmatic hernia and encephalocele.

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Conflicts of interest
The authors declare that there is no conflict of interest.

References