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CASE REPORT

Nasoethmoidal and Nasofrontal Encephalocele: case report

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Encephalocele,
Genetics,
Down Syndrome.

Abstract

Objectives: The article reports the case of encephalocele nasofrontal in an infant, relating to the genetic, syndromic and socioenvironmental factors, showing the importance of the health team being attentive to the findings, being a malformation, with a high mortality and morbidity rate and having few reports in the literature. **Case Report:** Infant, female, born at term, vaginal delivery, 3.570g, natural and coming from Uberaba. After birth, she was hospitalized for a congenital heart disease and Down syndrome, cardiac surgery was performed. In this period, the respiratory physiotherapist perceived the difficulty of introducing the nasal aspirate, and was sent to the otorhinol, without statistical findings, a cranial tomography was performed at 4 months, which diagnosed an infant with nasoethmoidal and nasofrontal encephalocele. The encephalocele is a rare anomaly, with high morbidity, mortality and risk of complications. It is related to other pathologies diagnosed in patients and to the antecedent factors at birth, genetic and environmental causes. **Comments:** Although it is difficult, with high mortality and morbidity, it is of great value to know about the factors that are related to this anomaly, both for the early diagnosis and for the prevention.

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INTRODUCTION

Encephalocele is a protrusion of skull content beyond its normal limits, through a congenital bone malformation, or more rarely, through normal skull foramina or fissures[1]. A relatively rare malformation, which occurs due to a defect in neural tube closing during the embryonic development phase, between the third and fourth week of gestation[2].

The overall prevalence of encephalocele is estimated at 1 case for every 35,000 to 40,000 live births. Primary encephalocele is divided into occipital, cranial vault, frontoethmoidal, and basal encephalomeningocele[3]. The basal forms, even rarer, constitute only 2% to 10% of these encephaloceles[4,5].

Nasoethmoidal or transethmoidal encephalocele is uncommon, and results from a congenital bone defect in the cribriform plate[6], usually located in the nasal cavity, close to the septum and medial to the middle turbinate[1]. Basal encephalocele is not directly visible and can cause upper airway obstruction[7].

The objective of this paper is to report a case of nasofrontal encephalocele, in a 4-month-old infant with Down Syndrome, which is a rare malformation, with a high rate of mortality and morbidity, and which has few reports in the literature.

In Brazil, there are few publications on the prevalence of Neural Tube Defects, 0.83:1000 to 1.87:100[8].

CASE REPORT

Patient M.S.M.B, white, female, 4 months old, born in Uberaba at term - at 38 weeks, by vaginal delivery, weighing 3,570 grams, daughter of a teenage mother - 14 years old. At birth, phenotypic changes and severe respiratory distress were identified, requiring ventilatory support. After complementary exams, Hypothyroidism, Down syndrome with congenital heart disease - total atrioventricular septal defect (AVSD), hypoplasia of the transverse arch and coarctation of the aorta with Chronic Heart Failure were identified, these findings were seen after birth, due to the lack of prenatal follow-up. She remained on ventilatory support for an extended period, with several attempts at extubation.

Since birth, she had respiratory difficulty and orotracheal intubation was performed. She remained intubated for three months with several extubation failures during this period. After the third month, extubation was possible, and she remained with the use of supplemental oxygen through a nasal catheter.

In one of the physical therapy assessments, there was difficulty during aspiration of the nasal cavity, and the suction tube found a block in her right nostril. Nasal endoscopy showed an intense inflammatory process, not being possible to access the right choana and external synechiae. The ENT advised on the intranasal use of dexamethasone, for such block could have been caused by an inflammatory process in her airway.

Even with the treatment, there was no improvement in the respiratory pattern, and a computed tomography of the sinuses showed bone defects in the skull base, in the ethmoidal cribriform plate and between the frontal and nasal bones, with signs of nasoethmoidal and nasofrontal encephalocele and apparent herniation of intracranial content to the nasal cavity (Figure 1) on the right and obliteration of ethmoid cells. The right side had pneumatized hypoplastic maxillary sinuses and to the left it was filled with mucous thickening, a common finding in this age group.

Due to the persistence of respiratory difficulty, we decided to perform a tracheostomy. One month after surgery, the infant presented respiratory distress, returning to mechanical ventilation. She evolved with a new infectious condition without response to treatment, presenting cardiorespiratory arrest on February 6, 2019.

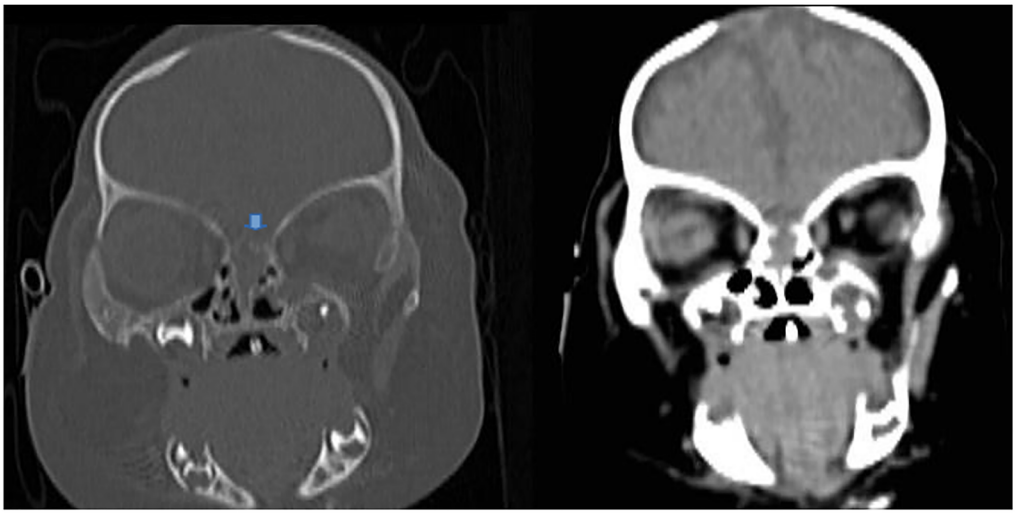


Figure 1. Paranasal sinuses CT scan, showing the encephalocele, with cranial contents herniating into the nasal cavity.

DISCUSSION

Neural tube closure defects (NTDs) have an incidence of 1-2 per 1,000 births, a condition resulting from failure of the neural tube to close between the third and fourth weeks of embryonic development[9,10]. The incidence of NTCD varies with age, race, geographic location, socioeconomic status and multiple predisposing factors, such as genetic disorder, chromosomal abnormalities and maternal risk factors such as diabetes mellitus, hyperthermia and exposure to teratogens[11].

There is evidence that NTCD cases are more frequent in the presence of other genetic syndromes[10].

A study carried out in a retrospective analysis of newborns admitted to the Neonatal Intensive Care Unit with NTCDs at the Security Forces Hospital, in Riyadh, Saudi Arabia, showed that of the 103 hospitalized newborns with NTDs, 20 (19.4%) had genetic, chromosomal and/or other underlying syndromic abnormalities[11].

In addition, there are reports in the literature that NTCDs are more common in females[12]. A study carried out with newborns diagnosed with encephalocele between 1997 and 2008, at Hospital da Mulher Prof. Dr José Aristodemo Pinotti, showed that there is a greater number of female children born with encephalocele (67.44%), with gestational age greater than or equal to 37 weeks (79.07%), and weighing more than 2500g (67.44 %)[13].

The Latin American Collaborative Study of Congenital Malformations (ECLAMC)[14] and another study[15] showed a higher prevalence of cases of neural tube closure defects in mothers younger than 19 years of age[14].

Another ECLAMC study, with a sample of 383 mothers with chronic disease, compared with 297 healthy mothers, showed that offspring of mothers with diabetes mellitus, bronchial asthma, hypertension or hypothyroidism have a higher risk of having a congenital malformation, when compared to offspring of healthy mothers[16].

Corroborating the studies cited above[10-16], in this case the infant was female, with congenital anomalies and the daughter of an adolescent mother, who also has hypothyroidism.

In addition to the rarity of the case, it is of great importance, for there is a high rate of morbidity, mortality and risk of serious complications. In addition, the healthcare team should consider and observe these findings.

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