Case Report

AN UNUSUAL CASE OF GIANT FRONTO-OCCIPITAL ENCEPHALOCELE

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ABSTRACT

An unusual case of large interparietal encephalocele is described in a newborn. In the present report, etiology, epidemiology, classification, clinical presentation and surgical approaches used for encephaloceles are discussed.

RESUME

Un cas inhabituel de large encéphalocèle de localization fronto-occipitale est décrit chez un nouveau-né. Dans la présente étude, nous discutons de l'étiologie, l'épidémiologie, la classification, la présentation clinique et l'approche chirurgicale utilisée pour la prise en charge de cette pathologie.

Mots-clés: Encephalocèle Géant – Localisation Fronto-Occipitale – Nouveau-né

INTRODUCTION

A two-day-old full term male infant born by cesarian section from non-consanguineous Rwandan parents. His mother, 36-year-old woman was admitted in the labor room for exaggerated fundal height and no progress of labor. At birth, he weighted 4 kg, his height was 50 cm and birth Apgar score was 6/10 then 8/10. The newborn presented a large fronto-occipital encephalocele characterized by protruding cystic swelling mass covering the whole head with slight discharge of liquid (Figure 1). Reflexes were decreased, and no other abnormality was found on his body. Unfortunately, he had cardiorespiratory arrest after 2 days of life while further radiological and brain CT-scan investigations were planed to be performed after transfer to a specialized referral hospital

Figure 1: Photograph showing a newborn with a giant fronto-occipital encephalocele

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DISCUSSION

Encephalocele is a type of neural tube defect that occurs very early in fetal life where the embryo's cells that form the skull do not come together to close over the brain. The result is a defect in bones of the skull, causing brain tissue to protrude from the skull. The protruding tissue may be located on any part of the head but is usually in the middle at the back of the head (midline occipital area). There may be meninges, brain tissue, parts of the ventricles and bone that protrude from the skull [1].

The exact etiology of this condition has not been determined by the medical experts yet.

As aforementioned, the neural tube fails to close completely during fetal development, causing sac-like protrusions of the meninges. These congenital abnormalities may arise due to a genetic cause. It is more common in families with a history of spina bifida and anencephaly, both of which are severe neurological disorders. Exposure to certain environmental substances before or during pregnancy may inhibit the proper development of the neural tube during the embryonic stage.

The incidence of encephalocele is 1 per 5000 live birth [2]. The most Common site of encephalocele is occipital (75%), followed by frontoethmoidal (13% to 15%), Parietal (10% to 12%) or sphenoidal [3].

However, the association of anterior (frontal) and posterior (occipital) involving parietal locations has not yet been reported in literature. Our patient presented a giant encephalocele combining both posterior and anterior locations but unfortunately he died before radiological investigations. Larger encephaloceles may be complicated by CSF leakage followed by infection [4]. In most cases, the condition is treated through surgical means. Patients with giant encephalocele and large amount of brain tissue in the sac

usually die either shortly after birth or as a result of operation. Surgery aims at putting the brain section outside the skull back into its original position, and closing the aperture (opening). In some cases, neurosurgeons even manage to repair large lumps of this type without further hampering the functioning ability of an affected child. Reparative surgery, usually performed during infancy, is presently the only effective treatment for this disease [5]. Ultimate surgical result depends on the amount of normal brain tissue left inside the skull after the operation. The ultimate prognosis depends on the extent and nature of herniated contents and associated anomalies and many large encephaloceles have an excellent prognosis despite their size.

Although prenatal detection may have an impact on the prevalence of encephaloceles among live births; a substantial number of infants continue to be born with encephaloceles. The risk of mortality for such infants is the highest during the first day of life, continuing to occur through adolescence and is influenced by several clinical and demographic characteristics including the site of defect, the contents of sac, the low birth weight, and associated congenital abnormalities. Ultrasound can detect the occipital encephalocele, and it is widely available. CT Scan is preferred for visualization of internal and external bony defects. MRI can visualize the herniated contents within the sac and help in detecting other brain anomalies [6].

CONCLUSION

Although the condition cannot be prevented completely, certain steps can be taken to decrease the chances of development. Proper intake of folic acid in the first few weeks of pregnancy may reduce the occurrence of this form of NTD. Folate is an important constituent in the synthesis and regulation of new cells required for DNA and RNA synthesis. It also plays an essential role in the nucleic acid synthesis and methylation. Physicians normally recommend pregnant women to consume 400 micrograms of folic acid daily. It is also important to refrain from smoking and drinking to ensure proper growth of the fetus. To the best of our knowledge, this is a first case report of giant encephalocele combining both frontal and occipital locations.

REFERENCES

1. C. Siffel, L. Y. Wong, R. S. Olney et al. "Survival of infants diagnosed with encephalocele in Atlanta, 1979-98," Paediatr Perinat Epidemiol, vol. 17, no. 1, pp. 40-8, Jan, 2003.
2. J. Mealey, Jr., A. J. Dzenitis, A. Hockey. "The prognosis of encephaloceles," J Neurosurg, vol. 32, no. 2, pp. 209-18, Feb, 1970.
3. D. G. McLone, "Congenital malformations of the central nervous system," Clin Neurosurg, vol. 47, pp. 346-77, 2000.
4. A. Aslan, O. Eser, O. Dogru et al. "Occipital mega encephalocele associated with acute inflammation," Pediatr Neurosurg, vol. 43, no. 1, pp. 65-6, 2007.
5. A. K. Mahapatra. "Giant encephalocele: a study of 14 patients," Pediatr Neurosurg, vol. 47, no. 6, pp. 406-11, 2011.
6. T. P. Naidich, N. R. Altman, B. H. Braffman et al. "Cephaloceles and related malformations," AJNR Am J Neuroradiol, vol. 13, no. 2, pp. 655-90, Mar-Apr, 1992.