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Giant Occipital Encephalocele: Case Report

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1. Abstract

Encephalocele is a malformation due to a neural tube closure defect associated with herniation of brain tissue and/or meninges through this congenital cranial defect. The size of an encephalocele varies from a few centimetres to a huge mass called a 'giant encephalocele'. Usually, the contents of the malformation are degenerative nervous tissue, meninges and a cystic part. Encephalocele is diagnosed prenatally by maternal screening of serum alpha-fetoprotein levels and by ultrasound. On two-dimensional ultrasound, encephalocele appears as a cystic mass with heterogeneous contents in continuity with certain cerebral structures. 2D ultrasound detects around 80% of encephalocele cases. Diagnosis is easy and reliable on the basis of ultrasound findings during the second trimester, and can also be made during the first trimester. The prognosis of newborns with encephalocele depends on the extent of herniation of neural tissue in the sac and the presence of associated anomalies.

2. Introduction

Encephalocele is a malformation caused by a defect in the closure of the neural tube, leading to herniation of brain tissue and/or meninges through this congenital cranial defect [1]. It is a rare anomaly occurring in 1 in 5,000 births worldwide, 70% of which occur in the occipital region [2]. The size of an encephalocele varies from a few centimetres to a huge mass called a 'giant encephalocele' [3]. Usually, the contents of the malformation are degenerative nervous tissue, meninges and a cystic part [4]. We report a case of occipital encephalocele associated with retrognatism in which we emphasise the importance of meticulous clinical assessment, the crucial role of brain imaging in diagnosis, and the possibility of multidisciplinary management (reconstructive neurosurgery, ophthalmology and paediatrics) which is often lacking in the local context of limited resources.

3. Patient

A new-born born to non-consanguineous parents, to a 29-year-old mother of IIG and IIP, with no particular pathological history and whose anamnesis found no signs of infection or medication taken during the pregnancy. She had received three antenatal counselling sessions, the first two in the second trimester and the last in the third trimester. She had received folic acid during the first trimester. A morphological ultrasound at 30 weeks revealed a large meningocele with extracerebral development communicating with the posterior cerebral fossa, cerebellar and vermin atrophy and supratentorial ventriculomegaly. An emergency caesarean section was performed early in labour. This resulted in the birth of a female neonate, weighing 2400 g, APGAR score 7/10, head circumference 33 cm with an anterior fontanel that was not bulging. The neurological examination was normal. She presented with an enormous giant occipital mass measuring 40 × 25 cm and 18 cm with positive transillumination. The rest of the external morphological examination was normal.

4. Discussion

The incidence of encephaloceles is 1 to 3 cases per 10,000 live births [2]. They are defined as a herniation of brain tissue and/or meninges outside the skull through a congenital bone defect due to a defect in the closure of the cranial part of the neural tube [2]. The aetiopathogenesis of encephaloceles is still controversial and several theories have been put forward, such as maternal hyperthermia, valproic acid, hypervitaminosis A, vitamin B12 and folic acid deficiency [5]. An important gene associated with occipital encephalocele is CEP290 (Acentrosomal Protein 290) [6]. Occipital encephalocele is frequently associated with neurological disorders, some infants may be asymptomatic on physical examination [7], but others may present with many different signs and symptoms such as delays in reaching developmental milestones, intellectual disability, learning disabilities, growth retardation, seizures, visual impairment, lack of coordination of voluntary movements (ataxia), hydrocephalus, spastic paraplegia or quadriplegia and microcephaly [8]. Antenatal ultrasound in trained hands is the examination of choice for antenatal screening for cerebral malformations. It can detect a cranial defect and sometimes a herniation of the brain. It shows a mass in the median line of the skull, more often in the occipital than the frontal region [9].Surgical repair of encephaloceles can be carried out safely when the technical conditions are met. The aim is to ensure a tight, physiological and cosmetic closure. The surgical approach varies, and the common pitfall is inadequate closure of the dura, leading to post-operative cerebrospinal fluid (CSF) leakage or the formation of a pseudomeningocele [1]. Early surgical treatment before significant craniofacial dysmorphia develops gives satisfactory cosmetic results [10].The preventive measures adopted in a number of developed countries, such as taking folic acid during the periconceptional period, antenatal diagnosis by imaging (ultrasound or MRI), legislation on therapeutic termination of pregnancy, etc., have all been taken into account.

5. Conclusion

Occipital encephalocele is the most common form of encephalocele and manifests itself as a congenital swelling of various sizes on the occipital bone in the midline. If a patient is found to have encephalocele, a search must be made for associated malformations, which in some cases constitute amniotic bridle syndrome. Diagnosis is based primarily on ultrasound. Surgery is the standard treatment from birth. Overall management begins with raising public awareness of the need for good pregnancy monitoring and, above all, strengthening preventive strategies through folic acid supplementation during the periconceptional period.

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