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A GIRL WITH AN ASYMPTOMATIC PARIETAL ENCEPHALOCELE

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Encephalocele is a general term that denotes protrusion of cranial contents beyond the normal border of the skull. A female infant with a soft swelling on the head diagnosed as posterior parietal encephalocele at the age of 8 months is presented. The reason for late diagnosis was most likely her asymptomatic presentation. Neurosurgical repair was simple, as the lesion containing only cerebrospinal fluid was small. The postoperative period was also uncomplicated. Nevertheless, the child was followed up later on. In addition, genetic analysis by high-resolution micro-array technique was found to be normal.

Descriptors: ENCEPHALOCELE; INFANT; NEUROSURGERY; GENETIC TESTING

INTRODUCTION

Encephalocele is a general term that denotes protrusion of cranial contents beyond the normal borders of the skull (1, 2). The term includes meningocele (herniation of the meninges with cerebrospinal fluid), encephalomeningocele (herniation of the meninges and brain tissue), and hydroencephalomeningocele (herniation of the meninges, brain tissue and part of the ventricular system).

The incidence of encephalocele is very low, in the range of 0.8 to 4 *per* 10.000 live births (3-5). However, the exact number is certainly larger as many encephaloceles result in miscarriage (6, 7). Six main groups of encephalocele have been proposed, according to the location of the defect in the cranium: occipital, occipitocervical, parietal, sincipital, basal, and temporal (8). Further classification within each group has also been suggested. The frequency and site of encephaloceles are geographically and racially very variable. Nasofrontal encephaloceles are much more common than posterior lesions in Southeast Asia (9). In North America and Europe, encephaloceles are less frequent and mostly occur in the occipital region (10). Most encephaloceles occur sporadically, with only a small proportion occurring as part of the recognized syndrome (4). Although all are congenital, some lesions may not be detected immediately after birth. The presentation is variable, regarding the region, size and contents of the encephalocele, and may vary from asymptomatic protrusion on the skull to very severe cranial disfigurement and mental retardation. The lack of other brain pathology and the absence of neural tissue within the sac carry a better prognosis for these lesions. After thorough diagnostic procedures, the treatment of choice is neurosurgical repair tailored to the presentation of the encephalocele (2, 8).

We present a girl with posterior encephalocele presenting as an asymptomatic protrusion of the head with meningocele containing only cerebrospinal fluid, diagnosed at 8 months. The encephalocele was located in the region of the posterior fontanel. Therefore, it was classified as a parietal posterior encephalocele.

PATIENT PRESENTATION

A female infant was born as the first child to healthy young unrelated parents. The labor was normal at 40 weeks of gestation. Birth weight was 3200 g, birth length 50 cm, head circumference 36 cm, and Apgar score 8/9. The patient was first examined in our neurosurgical outpatient clinic at the age of 8 months because of a soft swelling on the head. According to the parents, it was present since birth and emerged only slightly during the intervening period. The birth and the postpartum period were otherwise normal. She was vaccinated regularly, had no other diseases, and her development was normal. The family history for neurologic diseases was negative. The examination revealed no neurologic deficits or any other somatic abnormality apart from slight transient hirsutism in the pubic region that disappeared after several months. All hormonal tests were normal and so was abdominal as well as heart ultrasound. The head was of normal size and the anterior fontanel was closed. There was a soft midline protrusion in the region of the posterior fontanel measuring approximately 2 cm in diameter. The skin was angiomatous in this region and a tuft of hair was growing from the protrusion. No other abnormality of the head was observed. Skull x-ray revealed a small, round, midline skull defect with a hypersclerotic edge. Magnetic resonance imaging (MRI) revealed a midline encephalocele in the region of the posterior fontanel with a small stalk connecting the cele to the intracranial space.

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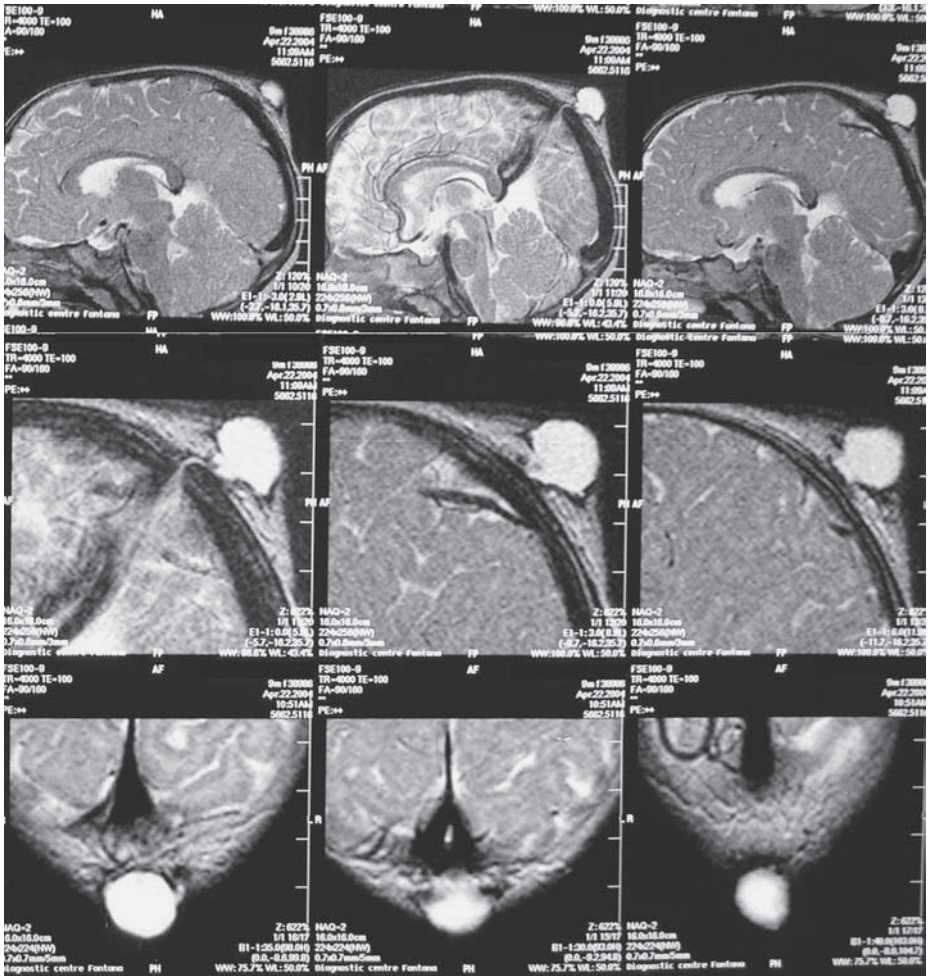

Figure 1. *Preoperative magnetic resonance scan showing parietal encephalocele in the region of the posterior fontanel, measuring approximately 2 cm in diameter, with a small stalk connecting the cele to the intracranial space. No other brain anomaly is seen and there are no signs of hydrocephalus (sagittal and axial views are shown).*

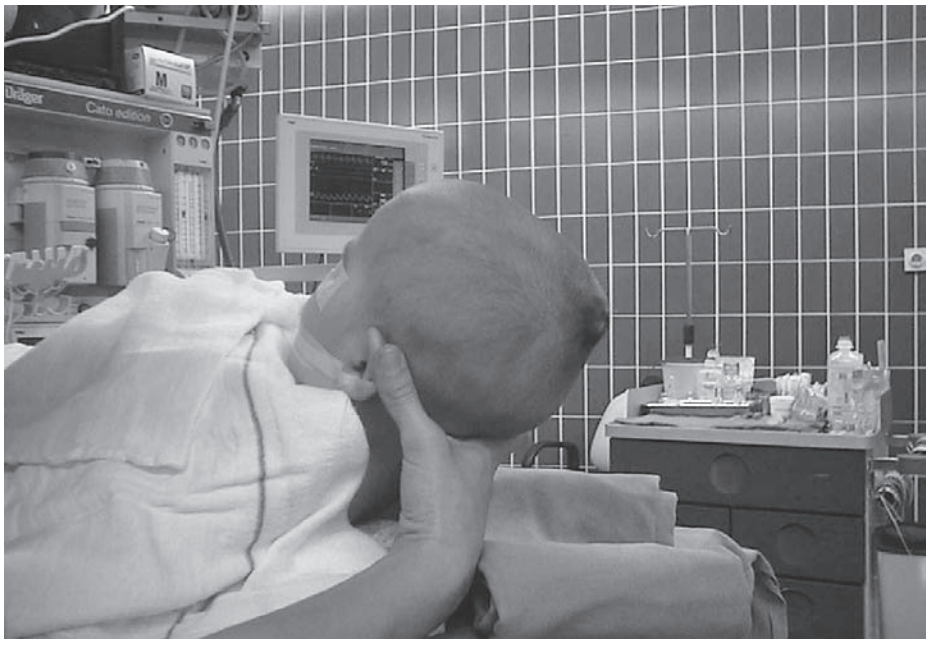

Figure 2. *Preparation of the patient for surgery. The encephalocele is visible as a protrusion in the posterior parietal region of the head.*

According to this, it was classified as a parietal encephalocele. The cele appeared to contain only cerebrospinal fluid and no neural tissue was seen. The encephalocele was atretic with only a small stalk extending intracranially. No other brain abnormality was seen on MRI nor were there any signs of hydrocephalus (Figure 1).

Surgery was performed at the age of 12 months. The patient was placed in the prone position with the head facing down (Figure 2). A horseshoe skin incision was performed, the skin flap was carefully raised, and the skull defect with the stalk leading to the cele was identified. The cele was then opened and its content inspected. It contained only cerebrospinal fluid. There was no neural tissue seen. A small craniotomy was performed around the bony defect and the stalk was freed all the way down to the dural entrance immediately lateral to the sagittal sinus. The stalk was then ligated and cut from the normal dura and the dural defect closed watertight. The cele and the excess skin above it were removed. The skin flap and the skin defect in the cele region were closed with single sutures.

The postoperative period was uncomplicated. The child had no neurologic deficits and regular head ultrasound showed no signs of hydrocephalus. She was discharged one week after surgery. The ventricular size was also normal on follow up MRI two months after surgery. In addition, there were no intracranial abnormalities noted after the surgical procedure (Figure 3).

Genetic analysis by high-resolution micro-array technique was performed at 8 years of age at the University Medical Centre Maribor (Laboratory of Medical Genetics). Genomic DNA was prepared from blood samples and analyzed with the SurePrint G3 Human CGH Microarray Kit, 8x60K for Agilent Technologies. The result of the genetic analysis showed a normal female: arr (1-22, X) X2 (11).

DISCUSSION

Patients with posterior encephaloceles can present with a variety of different associated brain malformations: brainstem distortion, cerebral abnormalities, posterior fossa cysts, elevation of the tentorium and dural venous sinuses, caudal displacement of the occipital and temporal lobes, dorsal cysts that communicate directly with the ventricular system, holoprosencephaly, split sagittal sinus, and anoma-

lies of the galenic vein (12, 13). Dorsal cysts and holoprosencephaly are more common with parietal encephalocele. For this reason, their prognosis is often worse than that of occipital encephalocele. Patients with a posterior encephalocele can also have extracranial malformations, the most frequent being cardiac anomalies, cystic kidneys, limb malformations and polydactyly (14). There was no other brain abnormality or extracranial malformation observed in our patient.

MRI is the imaging procedure of choice for evaluating encephaloceles and the contents of the sac, and for a reliable identification of its relationship to the neural and vascular structures (15). Computed tomography can be of assistance, especially in delineating bony defect. Digital subtraction angiography can offer more precise information regarding arterial and venous structures and their anomalous configuration. In our uncomplicated case, MRI offered enough information to enable proper planning of the neurosurgical procedure (1).

Atretic encephaloceles typically present as small, skin-covered midline subscalp masses or cysts containing meninges and/or neural rests, and are located in the parietal region in approximately half of the cases (16).

The pathogenesis of encephaloceles remains obscure. Various theories have been postulated: a defect in primary cranial neurulation, a disturbance in the separation of neural and surface ectoderm after neurulation, secondary herniation of the brain and meninges due to a developmental failure of ossification, and the theory of neural crest remnants. Most probably, various explanations hold true for different types of encephaloceles (4, 17). Genetics may also play a role in this malformation (18, 19). In our patient, the latter was normal.

Neurosurgical repair is simple for small lesions containing only cerebrospinal fluid but can be very demanding for large lesions containing important nervous and vascular structures. The aim of surgery is to remove the sac and to preserve functional neural tissue. For atretic encephaloceles with a stalk that extends intracranially, the stalk should be identified and divided at the level of the dura. Watertight closure of the dural defect is mandatory to prevent cerebrospinal fluid leakage and central nervous system infection. This procedure was also used in our case. For large encephaloceles, extensive reconstructive techniques must be employed (1).

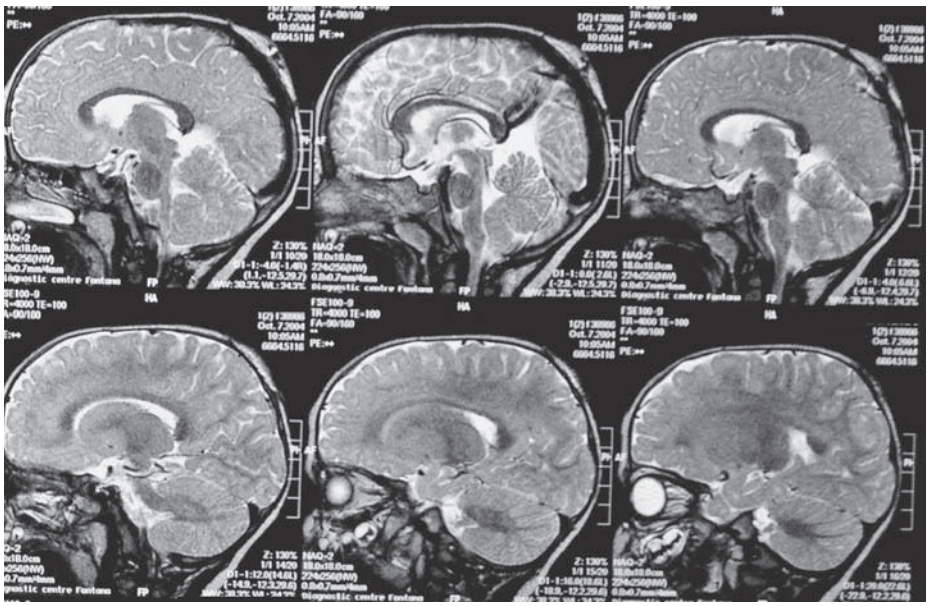

Figure 3. *Postoperative magnetic resonance scan showing normal brain structures in the region where surgery was performed, with no signs of hydrocephalus (sagittal view is shown).*

CONCLUSION

Encephalocele is a rare clinical entity with a variable presentation, clinical picture and sequele. With small lesions not containing neural tissue, the surgery is simple and safe and the prognosis mostly favorable. Close observation is necessary in the postoperative period to detect central nervous system infection or the development of hydrocephalus.

Authors declare no conflict of interest.
Autori izjavljuju da nisu bili u sukobu interesa.

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S a ž e t a k

DJEVOJČICA S ASIMPTOMATSKOM PARIJETALNOM ENCEFALOKELOM

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Encefalokela je općenit pojam koji označava proboj sadržaja lubanje izvan normalne granice lubanje. Prikazuje se slučaj ženskog dojenčeta s mekim otokom na glavi koji je dijagnosticiran kao stražnja parijetalna encefalokela u dobi od 8 mjeseci. Ovako kasna dijagnoza vjerojatno je bila uvjetovana asimptomatskom manifestacijom. Neurokirurški zahvat bio je jednostavan, jer je oštećenje bilo malo i sadržavalo je samo likvor. Poslijeoperacijski oporavak bio je uredan, ali je dijete ipak bilo praćeno. Uz to, genetska analiza provedena tehnikom mikro-array visoke rezolucije bila je uredna.

Deskriptori: ENCEFALOKELA; DOJENČE; NEUROKIRURGIJA; GENETSKA ANALIZA

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