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□ Case Report □

Two cases of congenital atretic encephalocele misdiagnosed as dermoid cyst

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Atretic cephalocele is a degenerative form of encephalocele, which is detected as a cystic mass in the head, primarily in infants. Its presentation and prognosis vary and depend on various factors, including the nature of the tissues within the cyst, other concomitant anomalies, the site of development, and the presence or absence of an embryonic straight sinus. We here report 2 cases of atretic encephalocele, that were transferred to our hospital because round tumors, misdiagnosed as dermoid cysts, were detected in their parietal lobes immediately after birth. On diagnostic and differential MRI, an embryonic straight sinus was detected while histochemical results indicated that the lesions contained cerebral tissues. Despite these structural anomalies, the two patients developed normally neurologically and no other anomalies were detected. We here discuss these two cases and present a review of the relevant literature. **(Korean J Pediatr 2006;49:1000-1004)**

Key Words : Atretic encephalocele, Embryology, Magnetic resonance imaging

Inroduction

Atretic encephalocele was reported by James and Lassmann for the first time in 1972 as meningocele manqué, and it was defined as a degenerative form of encephalocele[1]. Subsequently, it was variously designated as atretic, occult, abortive and rudimentary cephalocele, amongst other terms. Its prognosis varies and depends on a number of factors, including the nature of the tissues contained within the lesion, the presence or not of other concomitant anomalies, the site of development, and the presence or not of an embryonal straight sinus[2].

We treated two patients who were referred to our hospital because a round tumor was detected in the posterior area of their parietal lobes immediately after birth. Different imaging techniques, including various simple imagings of the cephalic area, ultrasound, CT and MRI were used, which somewhat confused the diagnosis of atretic cephalocele. However, in subsequent follow-ups, no other anomalies or neurological abnormalities were detected. Hence two cases are reported here together with a review of the relevant literature.

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Case report

Case 1

A 1-day-old male neonate presented with the presence of a round tumor in the posterior area of the parietal lobe. Immediately after birth, a round tumor was detected in the posterior area of the parietal lobe, ultrasound was performed, and transferred to our hospital.

The patient was 38 weeks and 6 days old and weighted 3,200 g. He was born by Cesarean section and presented no special features during the prenatal examination. The family history and the obstetrical history of the patient's mother were normal.

At the time of admission, he appeared to be healthy and to have normal vital signs; the pulse rate was 160/minutes, respiration rate was 50/minutes, and his temperature was 36.8℃. Although the overall appearance was normal, a tumor was detected during an examination of the cephalic area.

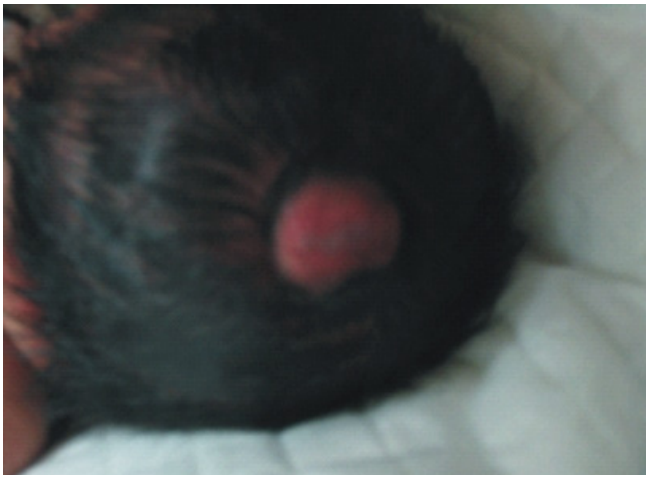

Fig. 1. The patient shows a circular cystic mass covered with scalp without hair at the parietal area.

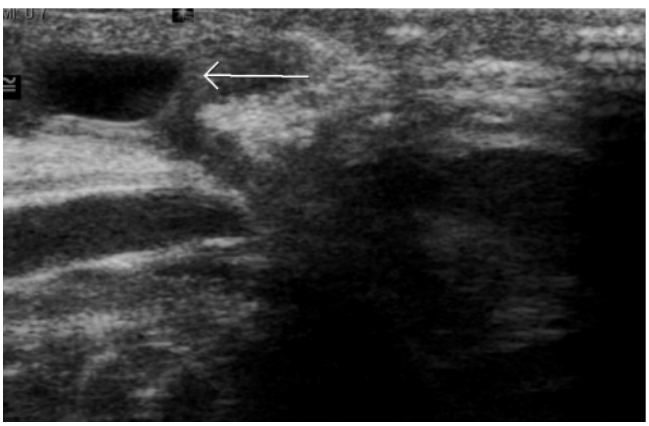

Fig. 2. USG shows 1.5 cm sized cystic lesion at occipital scalp with suspicious extension to intracranial portion.

The tumor was approximately 2×2 cm in size, elevated, covered with hairless skin and thus clearly demarcated from the adjacent scalp. When palpated, it was soft and did not move (Fig. 1). Upon further examination, the heart sound was regular, the lung sound was clean and abdominal examination did not reveal any special findings.

In blood test, hemoglobin was 17.5 g/dL, the total leukocyte number was 21,570/μL, platelet was 289×10^3/mm^3, blood glucose was 64 mg/dL, serum total protein was 5.5 g/L, albumin was 3.3 g/L, total bilirubin was 2.5 mg/dL, sodium was 136.1 mEq/L, potassium was 5.0 mEq/L, chloride was 109.2 mEq/L, and special results were not detected.

On simple images of the cephalic area, bone defect could be detected. However, a head ultrasound revealed a cystic lesion of approximately 1.5 cm in size in the parietal lobe. This appeared to be connected to the tissues contained in the cranial cavity. In the inside of cyst, low intensity findings were detected, which suggested that it was either a meningoencephalocele or dermoid cyst (Fig. 2).

On the 2nd day, despite performing cephalic CT, it was impossible to accurately evaluate the tumor, so cephalic MRI was performed. MRI revealed a cystic lesion 1.7 cm in size in the posterior area of the parietal lobe in the midline. Moreover, another small cyst was detected within the cyst that appeared to be connected to the tentorium within the cranium. Furthermore, the straight sinus did not take a normal pathway, but instead ran along the tentorium. It was directed towards the scalp of parietal lobe, and appeared to the connected to the side of superior sagittal sinus. On the basis of these observations the tumor was diagnosed as an atretic parietal cephalocele (Fig. 3). An ultrasound analysis

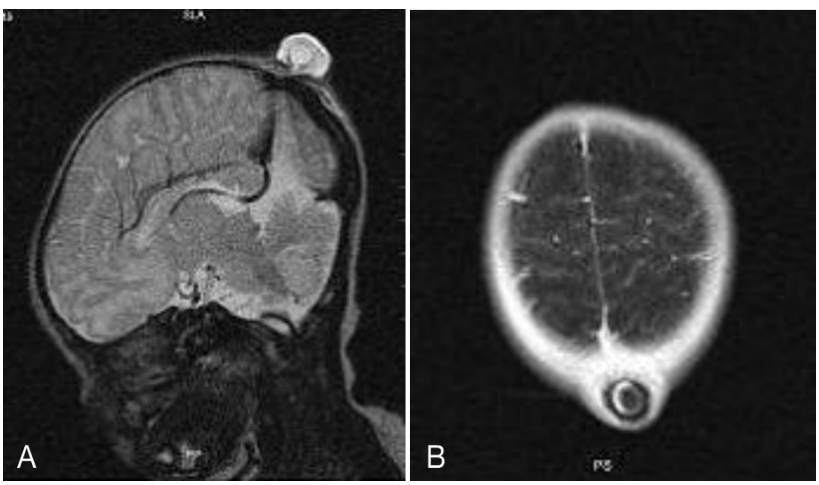

Fig. 3. Straight sinus and tentorium extends to the parietal scalp.

performed to evaluate spinal anomalies did not reveal any unusual features.

During the admission of the pediatric patient, the lesion showed the result to be increased and on the 5th day, surgery was performed by the department of neurosurgery. 1 day after surgery, oral nursing was initiated and during the follow up, the patient was discharged after finding that the nursing volume had increased and the body weight had improved. In subsequent outpatient follow-ups, no abnormal findings were detected.

On Surgical findings Dura mater tissue was detected in the cyst interior, whose content were mostly cerebrospinal fluid. Efforts were made to push tissues towards the inside of the cranium, but unfortunately this failed. Therefore, primary sutures were inserted after cyst resection.

Histologically the lesion was immunopositive for GFAP and was therefore diagnosed as an encephalocele (Fig. 4).

Case 2

A 20-day-old male neonate presented the presence of a mass in occipital area. Immediately after birth, a tumor was detected in the occipital area. The patient visited a private pediatric clinic and subsequently transferred to our hospital for specialized tests.

He was Born at 39 weeks by Cesarean section. No special features were detected during the prenatal examination. At the time of admission, his weight was 3,830 g. His vital signs were a pulse rate of 140/min, a respiration rate of 48/min, and a temperature of 37℃. On cephalic examination, a soft but fixed tumor of 2×3 cm was detected in the occipital area. No other abnormalities were detected during either chest and abdominal examinations.

Cephalic ultrasound releaved a small, low-intensith region in the occipital area, which appeared to be connected to the inside of cranium. A small tumor in the occipital area was detected by MRI (Fig. 5). Moreover, on MRI, the straight sinus appeared th be connected to the superior sagittal sinus (Fig. 5).

In follow-up examinations, this patient's tumor appeared to decrease in size. Thus, it was decided to discharge him and monitor him as an outpatient.

Discussion

Cephalocele is defined as the congenital extrusion of intracranial structures due to the defect of the closure of neural tubes. It develops primarily in the parietal lobe of the midline region of the occipital area, where it forms a small nodule or a cystic lesion generally covered by hairless scalp. In most cases, a palpable defect in the cranium is also present. Atretic cephalocele is classified by its cyst content, which generally include residual dura or nervous tissues. Among cephaloceles, the incidence of atretic cephalocele is reported to be 4-17%[3], of which parietal cephaloceles accounts for 37.5-50%[3, 4]. As these diagnostic designations are

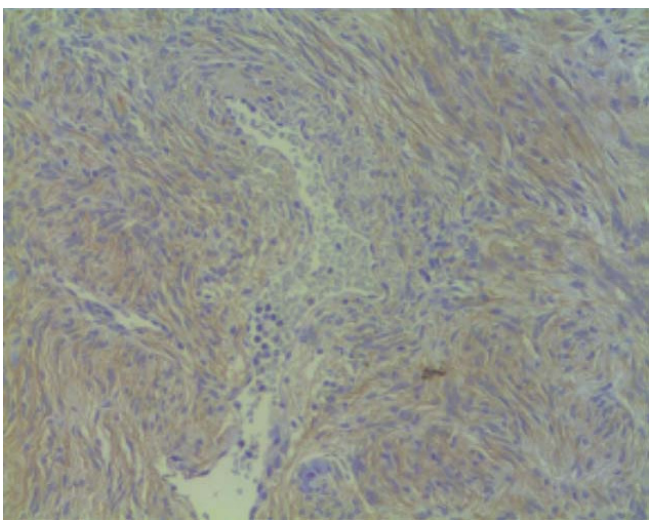

Fig. 4. Result of immunohistochemical stain has positive GFAP stain (×400).

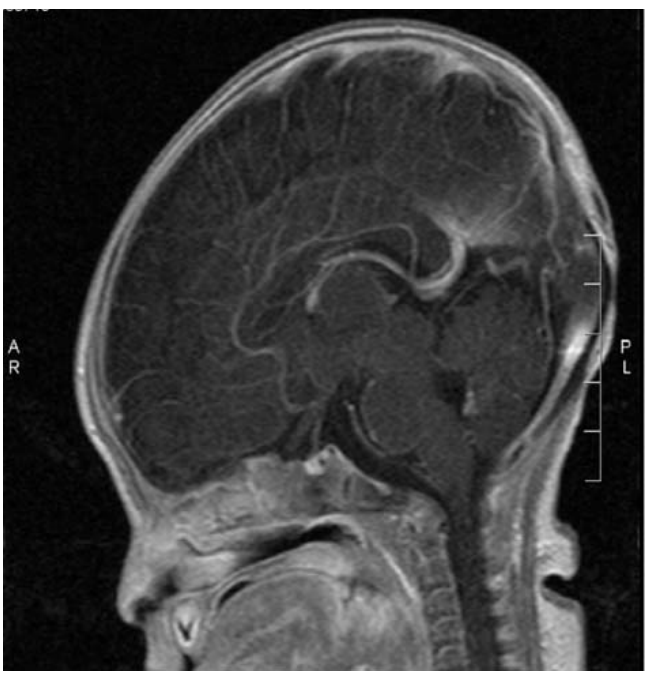

Fig. 5. Small subscalp mass in the lower mid-parietal area.

histopathological, the diagnosis is inferred based on clinical observations and images. In case reported here, the tumor was located in the median area and was covered with skin. Hence it was initially suspected to be a dermoid cyst or meningoencephalocele. However, the ultrasound analysis performed for the initial diagnosis revealed cystic tissue connected to the inside of the brain[5, 6]. On the 2nd day, an increase in size suggested that the cystic tissue was actually connected to the inside of the brain. Hence, cephalic CT was performed. However, since the cephalic CT view is limited sagittally, this was not particularly informative, and thus cephalic MRI was performed. MRI images revealed the presence of another structure within the cystic structure. In addition, a distinct connection to the inside of the brain and an embryonal straight sinus were observed. Hence, the lesion was deemed to be a cephalocele[7-9].

Richard et al.[10] subclassified cephalocele on the basis of whether an embryonic straight sinus is present or not. Moreover, embryonic positioning of the straight sinus plays an important role in marking the timing of an embryonic insult and in radiological diagnosis. In most cases the embryonic straight sinus is present and other concomitant anomalies are observed. While the absence of an embryonal straight sinus rare, its prognosis has been reported to be good. In our cases, the embryonal straight sinus was detected by MRI, which was very helpful in arriving at a diagnosis. However, in contrast to other reported cases, intracranial anomalies were not detected, and upon clinical examination, development appeared normal.

The prognosis of cephalocele varies depending on the site of its development, concomitant intracranial anomalies and various other factors[11]. In parietal lobe cases, which include dorsal cyst malformation, the lesion is generally accompanied by anomalies in the cranium and the eye, and most patients are related developmentally or die. On the other hand, occipital cephalocele, which is defined as a defect between lambda and bregma, is generally not associated with brain abnormalities. In cases of atretic cephalocele, it has been reported that occipital cephalocele is more common. In the cases reported here, the lesion was located in the posterior area of the parietal lobe, and in follow-up observations no other concomitant anomalies were observed. Recently, the importance of the location appears to have become less critical and it has been reported that generally the prognosis of atretic cephalocele itself is good[10].

The mechanism of cephalocele development is still controversia[12, 10, 13, 14]. Explanations include the incomplete resolution of a large meningoencephalocele formed during the early fetal period, the continuous presence of a fetal nuchal bleb and a neural crest trace. In this regard, tissues within cephaloceles are primarily heterotopic glial tissue, abnormal vascular tissues, and neural tissue remnants. However, in our first case, it was determined surgically that the interior of the cystic lesion was mostly filled with cerebrospinal fluid. Moreover, histological evaluation of the removed tissue revealed GFAP-positive brain tissues, which is consistent with it being a cephalocele.

Anomalies that often accompany cephalocele include hydrocephalus, intracranial cyst, cerebellar dysgenesis, low density of white matter and anomalous connection of the venous sinus. Aydin[15] and colleagues reported a case in which a cephalocele was connected to the lateral ventricle, resulting in an acceleration of its pulsation rate and an increase in its size while breathing. In addition, Martinez et al.[3] reported that in some cases with accompanying cerebro-oculomuscular syndrome and/or other complex cerebral anomalies, the presence of an atretic cephalocele is the sole clue that permits a correct diagnosis, and was described as the tip of the iceberg. In our cases, no outward anomalies were present, and no further anomalies were detected either on spinal ultrasound or durion surgery, suggesting a good overall prognosis.

The primary treatment strategy for cephalocele is surgery and postsurgical complications are rare. In one of our cases, surgical removal was performed without postsurgical complications. However, in the second case, the size of the lesion decreased gradually by itself and thus the patient was monitored by outpatient follow-up.

In conclusion, in infants that present a cystic mass in the parietal lobe, it is important to differentiate between lipoma, dermoid cyst, sinus pericranii, cephalocele. After clinical examination and neurological test, radiological tests should be performed, with MRI being the methodology of choice for rapid diagnosis.

한 글 요 약

유피낭종으로 오인된 atretic encephalocele 2례

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Atretic encephalocele은 1972년 James와 Lassmann에 의해 meningocele manque로 처음 보고 되었고 뇌류의 퇴행성 형태로 정의하였다. 이후로 atretic, occult, abortive, rudimentary cephalocele 등의 다양한 명칭으로 명명되었으며 포함된 조직, 동반 기형, 발생 위치, 배아성 직정맥동의 유무 등에 따른 차이점들에 대해 다양하게 보고 되어왔다. 두정엽의 후부에 낭성 종물을 주소로 입원한 환아의 진단 과정에서 두부 초음파, CT, MRI를 시행하였으며 그 과정에서 MRI가 진단에 가장 효과적인 영상을 제공하였다. 환아 진단 후 수술적 방법으로 종물을 제거하였으며 수술 중 이상 소견 및 추후 관찰 기간 동안 이상 소견 관찰되지 않았다. Atretic encephalcele의 희귀함과 진단 방법의 장단점 및 위치상 특성과는 다르게 동반 기형 및 발달 장애가 관찰되지 않은 증례를 치험 하였기에 보고하는 바이다.

References

1) James CC, Lassman LP. Spinal dysraphisms; spinal bifida occulta. London : Burrerworths. 1972;89-97.
2) Tomosato Y, Takao E, Masahiro I, Tadao N. Atretic cephalocele-report of two cases with special reference to embryology. Child's Nerv Syst 2001;17:674-8.
3) Martinez-Lage JF, Sola J, Casas C, Poza M, Almagro J, Girona DG. Atretic cephalocele : the tip of iceberg. J Neurosurg 1992;77:230-5.
4) Yokota A, Kajiwara H, Kohchi M, Fuwa I, Wada H. Parietal cephalocele: clinical importance of its atretic form and associated malformation. J Neurosurg 1988;69:545-51.
5) Francis B, Jeanine B, Dominique R, Dominique T, Isabelle S, Pascale S, et al. Intracranial venous anomalies associated with atretic cephaloceles. Pediatr Radiol 2000;30:743-7.
6) Abubacker S, Lakhkar B, Shetty D, Krishna G. Adult atretic parietal cephalocele. Neurol India 2002;50:334-6.
7) Otsubu Y, Sato H, Sato N, Ito H. Cephaloceles and abnormal venous drainage. Child's Nerv Syst 1999;15:329-32.
8) Gulati K, Phadke RV, Kumar R, Gupta RK. Atretic cephalocele : contribution of magnetic resonance imaging in preoperative diagnosis. Pediatr Neurosurg 2000;33:208-10.
9) Curnes JT, Oakes WJ. Parietal cephaloceles : radiographic and magnetic resonance imaging evaluation. Pediatr Neurosci 1998;14:71-6.
10) Patterson RJ, Egelhoff JC, Corne KR, Ball WS Jr. Atretic parietal cephaloceles revisited : an enlarging clinical and imaging spectrum? Am J Neuroradiol 1998;19:791-5.
11) Saatci I, Yelgec S, Aydin K, Akalan N. An atretic parietal cephalocele associated with multiple intracranial and eye anomalies. Neuroradiol 1998;40:812-5.
12) Martinez-Lage JF, Poza M, Sola J, Soler CL, Montalvo CG, Domingo R, et al. The child with a cephalocele : etiology, neuroimaging, and outcomes. Childs Nerv Syst 1996;12:540-50.
13) Hong EK, Kim NH, Lee JD. Atretic encephalocele/myelocele : case reports with emphasis on pathogenesis. J Korean Med Sci 1996;4:364-8.
14) Alexander GB, David M, Robin B, John AK. Autosomal dominant occipital cephalcele. Neurol 2004;62:1888-90.
15) Aydin MD. Atretic cephalocele communicating with lateral ventricles. Child's Nerv Syst 2001;17:679-80.s