Journal of Korean Medical Science
Vol. 11, No. 4, 364-368 Aug. 1996

Atretic Encephalocele / Myelocele

- Case Reports with Emphasis on Pathogenesis -

Eun Kyung Hong, M.D., Nam Hoon Kim, M.D. Jung Dal Lee, M.D.

Department of Pathology, College of Medicine, Hanyang University

Atretic encephaloceles or myelomeningoceles are frequently solid due to hamartomatous proliferation of fibrous tissue and blood vessels. Because of the fibrous nature of the tumor with no cystic cavity and unusual location with no connection to CNS, they are frequently regarded as insignificant hamartomas. Apart from this terminology, they are also described as cutaneous meningiomas or hamartomas with ectopic meningothelial elements by the presence of meningothelial cells. We report a case of atretic encephalocele in the parietal scalp of an 8 year-old boy and a case of myelomeningocele in the posterior mediastinum of a 31 year-old woman. The terms atretic encephalocele and myelomeningocele are more appropriate for these cases because they include their pathogenesis and the non-neoplastic nature of the lesion.

Key Words : *Atretic encephalocele, Atretic myelomeningocele, Cutaneous meningioma, Hamartoma, Pathogenesis*

INTRODUCTION

Encephalomeningocele(encephalocele) or myelomeningocele(myelocele) has been defined as herniation of the brain or spinal cord, respectively, together with its meningeal coverings through a congenital bony defect (Burger et al., 1991). Direct communication with intracranial or intraspinal cavity is the rule. However, cases having no direct communication or pin-point defect are encountered very rarely. Furthermore, if the cystic lesions are filled with overgrowing mesenchymal and neuroglial tissue, the diagnosis of encephalocele or myelocele is extremely difficult to make, either clinically or pathologically. This entity has been variously called atretic or rudimentary encephalocele or myelocele, cutaneous meningioma, and hamartoma with ectopic meningothelial elements, etc(Lopez et al., 1974 ; Yokota et al., 1988 ; Suster and Rosai, 1990 ; Martinez-Lage et al., 1992).

We report a case of atretic encephalocele and a case of myelocele, both presented with soft tissue masses, and discuss the pathogenesis with review of the literature.

Address for correspondence : *Eun Kyung Hong, M.D., Department of Pathology, College of Medicine, Hanyang University, Seoul 133-792, Korea*
Tel : (02) 290-8960/8252, Fax : (02) 296-7502

CASE REPORT

Case 1

An 8 year-old boy was admitted for a small scalp mass present since birth. The boy was born with an enlarged head. The head circumference at the time of presentation was 53.2 cm. It corresponded to 85~90th percentile for his age. The scalp mass was soft and yielding in consistency and had atrophic and alopecic overlying skin.

The brain MRI revealed severe hydrocephalus but not progressive at that time. A small cranial defect at the posterior midline of parietal area was noted, which was connected to the scalp mass(Fig.1). The connection between dermal mass and cerebrum was not evident.

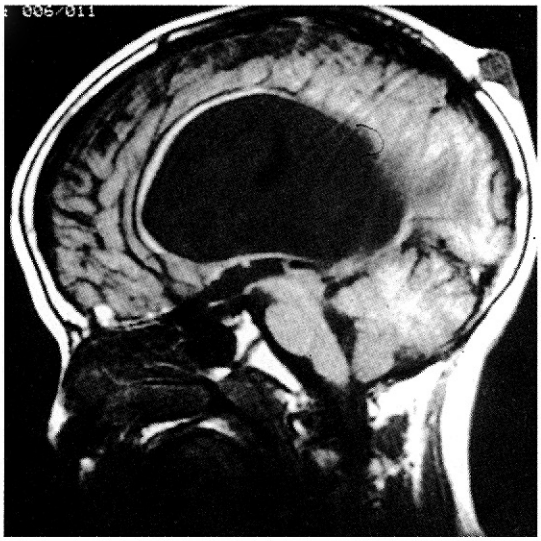

Fig. 1. Midline parietal location of dermal mass and associated hydrocephalus. A small bony defect is present beneath the dermal mass

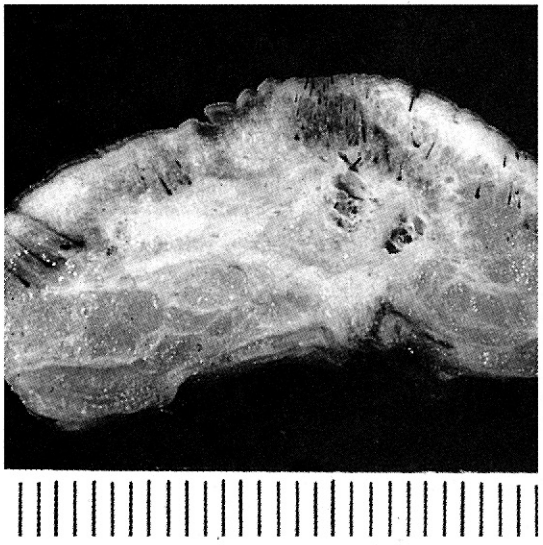

Fig. 2. Cut surface of protruding dermal mass on the scalp. The mass is solid and fibrotic.

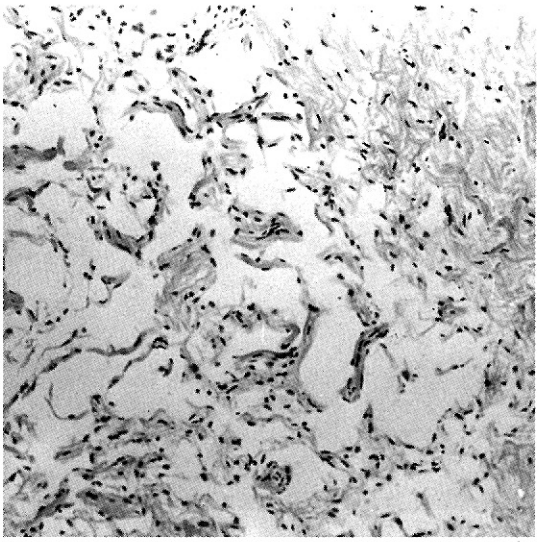

Fig. 3. Interanastomosing vessel-like channels in loose area.

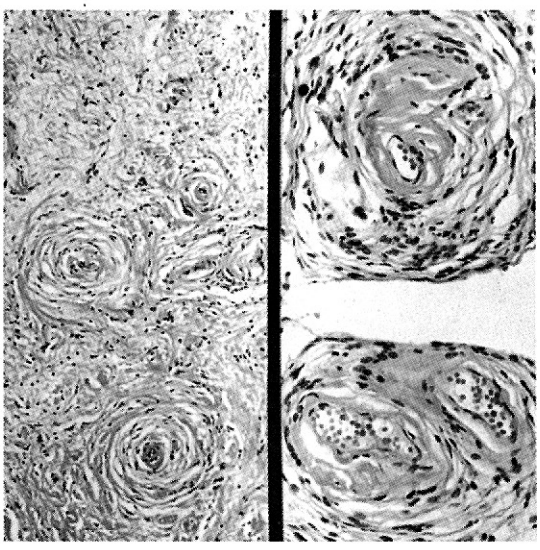

Fig. 4. Meningioma-like concentric whorls in fibrous background.

The mass was easily removed with overlying skin, and the surgeon found a blood vessel through this pin-point defect after the removal of the mass.

The excised mass was a relatively well defined, protruding fibrous mass of 2×1.5cm in cross diameter. The mass was gray white, solid with a few tiny microcystic cavities in the center(Fig. 2). Microscopic examination revealed proliferation of fibrocollagenous tissue alternated with loose edematous area. These wavy collagenous fibers were splitted by slit-like spaces lined by somewhat plump and elongated cells. They resembled vessel-like channels. Interanastomosing vascular

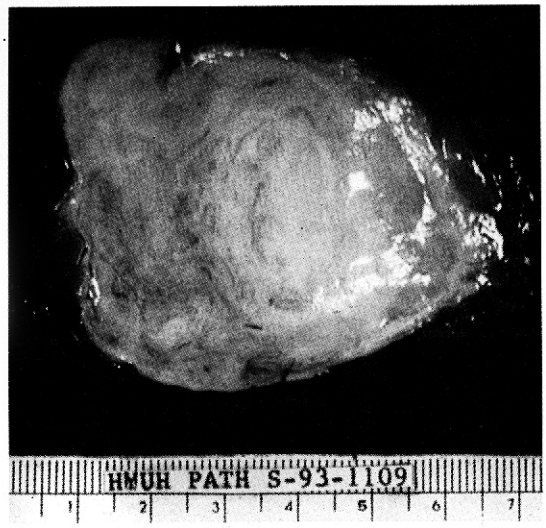

Fig. 5. Silvery white and homogeneously firm fibrotic tumor in the thorax.

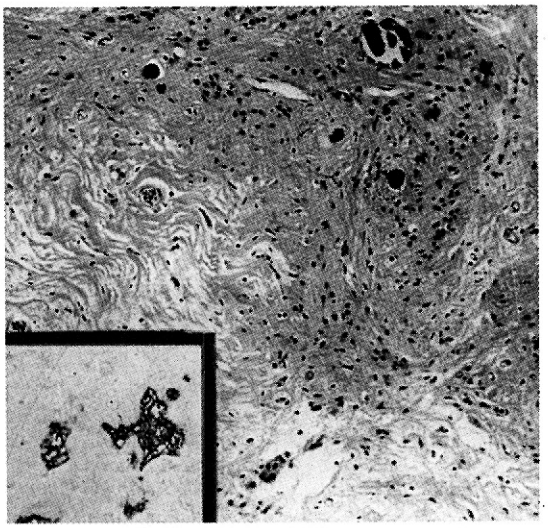

Fig. 6. GFAP(+) glial elements are admixed with wavy collagenous fibers.(Inset: GFAP stain).

channels were formed especially in loose edematous areas(Fig. 3). The overlying papillary dermis showed hemangioma-like aggregation of thin capillaries and absence of skin appendages. In areas, the whorling appearance of plump epithelioid cells and collagen fibers around vasculatures and even psammomatous calcification were noted in the center, like that of meningioma(Fig. 4). In a single focus, there was an island of neuroglial tissue within this fibrous proliferation.

Case 2

A 31 year-old female was admitted to the department of Internal Medicine for abdominal pain. During the work-up, a huge posterior mediastinal mass was incidentally found. The patient was previously healthy and showed no other abnormalities on physical examination and in laboratory work-up. Chest X-ray and CT scan revealed a paravertebral soft tissue mass with homogeneous density involving the level from T_8 to T_{11}. Under the impression of ganglioneuroma, the mass was removed. A thick fibrous cord was attached to the mass, which was connected to the abdominal sympathetic chain. No bony defect or abnormalities of the vertebra were found.

The excised mass was a well encapsulated, knobby, rubbery firm mass, measuring 6.5×5×2.5cm and weighing 45gm. A fibrous cord was attached to this tumor. The cut surface showed the typical silvery white whorled appearance of fibrous tumor. No cystic cavity was noted(Fig. 5).

Microscopic examination revealed the tumor was composed of dense collagen fibers, in which numerous foci of fine fibrillary neuroglial islands were intermixed (Fig. 6). Whorling of plump, epithelioid cells and occasional psammoma bodies were present, reminiscent of miningioma. There was also slit-like spaces lined by the same plump epithelioid cells resembling vascular channels. Immunohistochemical stains showed positive reaction to GFAP and NSE in neuroglial tissue, and positive reaction to EMA and vimentin in plump epithelioid cells.

DISCUSSION

Meningoceles or encephaloceles/myeloceles are cystic or saccular outpouchings with herniation of meninges, without or with neuroglial tissue, accompanied by underlying bony defects. Their development is usually explained by the failure of the neural tube to close or its subsequent rupture(Burger et al., 1991). In 1972, James and Lassman introduced an abortive form of meningomyelocele when this malformation was spontaneously arrested and did not progress during its development. There may be no underlying bony defect, but a bony sinus tract with a fibrous stalk connected to the dura mater or to the sagittal sinus is usually found on operation(Yokota et al., 1988; Martinez-Lage et al., 1992). This malformed lesion may be cystic, like conventional meningocele or encephaloceles/myeloceles.

However, most of the lesions show proliferation of dermal fibrocollagenous tissue and blood vessels as well as hyperplasia of arachnoid cell rests, forming solid, insignificant hamartomatous lesion in the subcutaneous tissue(Lopez et al., 1974). Many authors have applied the adjectives "atretic", "abortive", "occult", or "rudimentary" to this form of meningocele(Martinez-Lage et al., 1992).

This entity is morphologically quite distinct from more common conventional meningocele. It can be considered as an insignificant dermal nodule or hamartomatous lesion pathologically or clinically, if meningeal or glial elements were unrecognized(Schlitt et al., 1989; Drapkin, 1990; Martinez-Lage et al., 1992).

Suster and Rosai(1990) reported 5 cases of dermal hamartoma of the scalp with ectopic meningothelial elements. Histologically, there was an admixture of mature adipose tissue, abnormally arranged blood vessels, fibrous elements, and clusters of plump, cuboidal meningothelial cells. They stressed that the presence of interanastomosing vessel-like channels lined by plump epithelioid cells was a prominent feature and that differentiation from angiosarcoma was important. All five cases they reported had no bony defect or any other connection to the intracranial portion. However, these morphologic findings do not differ from so-called atretic meningocele or our present cases except that there was no glial element and that the meningeal component was minor or insignificant. They interpreted these lesions as hamartoma with an ectopic component of meningothelial cells arising on the basis of a developmental defect.

This dermal nodule was also regarded as a form of cutaneous meningioma(Lopez et al., 1974; Nochomovitz et al., 1985; Sibley and Cooper, 1989). Lopez et al. (1974) classified cutaneous masses into three groups when meningothelial cells were found ectopically within the mass. In these three groups, type I was primary cutaneous meningioma which was related to the developmental defects, and usually occurred in the scalp, face or paravertebral region of children and young adults. They suggested that this tumor probably originated from arachnoid cell rests displaced during embryogenesis into the cutis or subcutis. The development of this lesion was very similar to that of meningocele. The authors also described borderline cases between meningocele and cutaneous meningioma. According to the degree of their cystic nature and of fibrocollagenous proliferation, the borderline lesion was named as latent or rudimentary meningocele or as acoelic meningeal hamartoma.

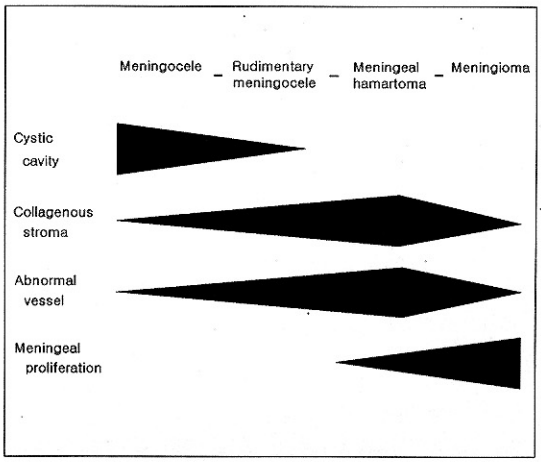

Fig. 7. Morphologic variations evolving from meningocele to cutaneous meningioma.

Thus these lesions can be categorized to the atretic or abortive meningocele on their pathogenetic point of view and to a distinct hamartomatous lesion or even meningioma on their morphologic point of view. They share the same pathogenetic mechanism with conventional meningocele. By the proportion of the components of these lesions, at least four categories of evolving pathology can be classified(Fig. 7)(Lopez et al., 1974). The two extremes are that of meningocele and of cutaneous meningioma. If the cystic cavity persisted and contained cerebrospinal fluid due to large bony defect, conventional meningocele or encephalocele/myelocele would be formed. When the cranial defect is small, after herniation of meningeal and/or neuroglial tissue, the defect may be restored spontaneously in utero. So the meningoceles once formed would be involuted or atretic(Yokota et al., 1989). By the presence of ectopic meningothelial cells, mesenchymal components of the soft tissue were stimulated and proliferated, replacing cystic cavity. Genuine neoplastic growth of this ectopic meningothelial cells could occur in case of cutaneous meningioma.

We prefer the term atretic encephalocele/myelocele because they reveal the non-neoplastic nature of the lesion as well as their pathogenesis of congenital, developmental abnormality.

Martinez-Lage et al(1992) classified atretic cephalocele into two types. Type 1 consisted of arachnoid tissue with clusters of anomalous blood vessels limited to the stalk of the lesion. It had intact, hair-bearing skin. Type 2 consisted of meningeal tissue intermingled with

dermal fibrous tissue, as well as clusters of anomalous blood vessels and ectopic neural or glial elements. The lesion extended beyond the stalk of the lesion. Our two cases can be classified as type 2 by this criteria.

Atretic cephalocele or myelocele itself is usually an unimportant and clinically benign lesion. The location of atretic cephalocele or myelocele does not differ from that of conventional cephalocele or myelocele. However, when it occurred in the parietal or occipital area, there may be other brain anomalies not pathogenetically related to the cephalocele or myelocele. There were some contradictory results of these associated anomalies. McLaurin(1964) and Yokota et al(1988) stated that parietal cephalocele were frequently associated with other brain malformations and carried much less favorable prognosis than those of the occipital region. In contrast, Martinez-Lage et al(1992) stated that the occipital location of cephalocele was associated with worse prognosis. More data will be needed to clarify the relationship between location and prognosis. Our case 1 was in the parietal location and was associated with arrested hydrocephalus. No other anomalies were found in this case.

The incidene of atretic cephaloceles is 4–17 % of all cephaloceles(Yokota et al., 1989), and the parietal location is not uncommon, comprising 37.5 %(Yokota et al., 1989) to 50 %(Martinez-Lage et al., 1992) of cases. When a scalp mass is noted at midline or near the vertex, the possibility of encephalocele should be ruled out and thorough neurological examination should be followed.

Our second case is also of note in terms of location. Intrathoracic meningocele is rare in incidence and there have only been slightly more than 100 cases reported (Auge and George, 1993). There is no mention of atresia in these reported cases. Because sixty five percent of reported cases have generalized neurofibromatosis and 52% have kyphoscoliosis, the development of intrathoracic meningocele was interpreted from a somewhat different point of view. The pathogenesis of intrathoacic meningocele is still unknown and there are two main hypotheses. One is primary regional bony defect or vertebral dysplasia and the second is primary dural dysplasia. Our presented case did not show any vertebral anomaly nor showed stigmata of neurofibromatosis. Only one fifth of reported cases had neither neurofibromatosis nor kyphoscoliosis(Auge and George, 1993).

We reported two cases of atretic or acoelic encephalo/myelomeningocele presented in unusual locations. Because of overgrowth of fibrocollagenous tissue and of unusual location, the lesion may frequently be misdiagnosed as an insignificant hamartoma. Detection of neuroglial tissue and concentric whorling arrangement of meningothelial cells is important not to miss this entity.

REFERENCES

Auge D, George R. *Intrathoracic meningocele. J Thorac Cardiovasc Surg 1993 ; 105 : 561-2.*

Burger PC, Scheithauer BW, Vogel FS. *Skull and related soft tissue. In Surgical Pathology of the Nervous System and its Coverings, 3rd ed. New York : Churchill Livingstone, 1991 ; 50-4.*

Drapkin AJ. *Rudimentary cephalocele or neural crest remnant ? Neurosurgery 1990 ; 26 : 667-74.*

James CC, Lassman LP. *Spinal dysraphisms : Spinal bifida occulta. London : Butterworths. 1972 ; 89-97.*

Lopez DA, Silvers DN, Helwig EB. *Cutaneous meningiomas-a clinicopathologic study. Cancer 1974 ; 34 : 728-44.*

Martinez-Lage JF, Sola J, Casas C, Poza M, Almagro MJ, Girona DG. *Atretic cephalocele : the tip of the iceberg. J Neurosurg 1992 ; 77 : 230-5.*

McLaurin RL. Parietal cephaloceles. *Neurology 1964 ; 14 : 764-72.*

Nochomovitz LE, Jannotta F, Orenstein JM. *Meningioma of the scalp : Light and electrom microscopic observations. Arch Pathol Lab Med 1985 ; 109 : 92-5.*

Schlitt M, Williams JP, Bastian FO, Smith S. *The small midline occipital encephalomeningocele : Definition of a syndrome. Neurosurgery 1989 ; 24 : 613-6.*

Sibley DA, Cooper PH. *Rudimentary meningocele : a variant of "primary cutaneous meningioma". J Cutan Pathol 1989 ; 16 : 72-80.*

Suster S, Rosai J. *Hamartoma of the scalp with ectopic meningothelial elements : A distinctive benign soft tissue lesion that may simulate angiosarcoma. Am J Surg Pathol 1990 ; 14 : 1-11.*

Yokota A, Kajiwara H, Kohchi M, Fuwa I, Wada H. *Parietal cephalocele : Clinical importance of its atretic form and associated malformations. J Neurosurg 1988; 69: 545-51.*