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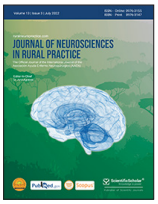

Case Series

Atretic cephaloceles with different imaging phenotypes – Case series with review of literature

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ABSTRACT

Atretic cephaloceles (ACs) are congenital skull defects with herniation of rudimentary intracranial structures through the defect and associated with persistent falcine sinus or embryonic positioning of straight sinus. We describe five cases of ACs, out of which only one had embryonic straight sinus. Three cases had other intracranial malformations such as hypoplasia of corpus callosum, dysplastic tectum in one child and parieto-occipital polymicrogyria with falcotentorial dehiscence in the other, and frontal horn deformity and cortical dysplasia in the third. The prognosis of AC depends on the coexistent intracranial abnormalities and this highlights the role of magnetic resonance imaging in diagnosing the other associated anomalies for prediction of prognosis and planning of necessary surgical management.

Keywords: Atretic cephalocele, Magnetic resonance imaging, Intracranial malformations, Persistent falcine sinus, Embryonic straight sinus

INTRODUCTION

Cephalocele is the external protrusion of the central nervous system contents through a defect in the cranium, most of which are midline in position. Atretic cephalocele (AC), also referred to as occult or abortive or rudimentary cephalocele, was first reported in 1972 by Martinez-Lage.[1] It is a midline, subscalp lesion covered by skin and consists of meninges and/or neuronal or glial cells. Cephaloceles can occur in occipital, parietal, frontal, petrous apex, or intrasphenoidal regions. Of cephaloceles, the incidence of AC is reported to be 4–17%, of which parietal 50–55% and occipital about 37%.[1-3] We report five cases of ACs, of which three are in occipital region, one each in the frontal and parietal region with a brief review of the literature.

CASE DETAILS

The age group of these children varied from 2 months to 14 years and presented with scalp swelling since birth. The third child had associated bilateral sensorineural hearing loss. No other deficits were noted in other children. All of them underwent magnetic resonance imaging under sedation and with intravenous contrast.

There were three occipital ACs – one at Torcula level, other two suboccipital – and one each in left frontal and in parieto-occpital region at lambda above the Torucla. The skull defect was small (3–11 mm) and the AC measured from 1 to 2 cm in size. Except the third child, no herniation of cerebrospinal fluid (CSF) or brain parenchyma was noted. No CSF tract was noted along the interhemispheric fissure/falx or tentorium or through the venous sinuses.

The first child showed dysplastic tectal plate and partial aqueductal obstruction with posterior corpus callosal hypoplasia. The third child with left paramedian suboccipital AC showed mild prominence of central canal of cervical spinal cord. The left frontal AC was associated with crowding of gyri and abnormal sulcus coursing in the left basifrontal region without reaching the frontal horn of the lateral ventricle, possibly incomplete schizencephalic cleft with cortical dysplasia. The last child apart from hypoplasia of posterior corpus callosum and septum showed parieto-occipital polymicrogyria, falcotentorial dehiscence, and prominent CSF spaces. The clinical and imaging features of the cases are summarized in [Table 1 and Figure 1].

DISCUSSION

The various theories for the etiology of ACs proposed are (i) failure of neural tube closure, (ii) reopened neural tube, (iii) neural crest remnant, (iv) nuchal bleb persistence, and

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Table 1: Clinical and imaging features of five cases of atretic cephaloceles.

	1 [Figure 1]	2	3	4 [Figure 2]	5 [Figure 3]
Age/gender	12 Y/M	2 Y/F	5 Y/M	14 Y/F	2 months/M
Location and relation to Torcula	Occipital at Torcula level	Suboccipital midline, below the Torcula	Suboccipital left paramedian, below the Torcula	Left frontal	Parieto-occipital, above the Torcula
Size of calvarial defect and cephalocle	1 cm 1.5 cm	2–3 mm 1.9 cm	3 mm 2 cm	3 cm 1.2 cm	1.1 cm 1 cm
Other abnormalities	1. Thinning of posterior body and splenium of corpus callosum 2. Dysplastic tectum with partial aqueductal obstruction	Nil	Prominent central canal in cervical cord	1. Prominence and lateral deviation of frontal horn of left lateral ventricle 2. Left frontal cortical dysplasia with deep sulcus	1. Diffuse thinning of corpus callosum with absent posterior septum and dilated lateral ventricles 2. Posterior falx dehiscence with wide parieto-occipital CSF spaces 3. Parieto occipital polymicrogyria
Straight sinus	Normal	Normal	Normal	Normal	Replaced by long midline vein to reach Torcula
Falcine sinus	Not present	Not present	Not present	Not present	Present
CSF tract	No	No	Small blind tract	No	No

CSF: Cerebrospinal fluid

(v) sequelae of primary mesenchymal injury.[3] Yokota *et al.*[3] in 1988 differentiated two types of ACs based on location and histopathological features – first parietal cystic type with fibrous content without neural or dermal tissue and dural connection, second occipital nodular lesions with fibrous core with dural connection at the level of torcula. The parietal type was more associated with other intracranial malformations such as hydrocephalus, intracranial cyst, cerebellar dysgenesis, anomalous venous drainage, white matter changes, holoprosencephaly, gray matter heterotropia, and Chiari malformations.

Martinez-Lage *et al.*[1] classified them into two groups, namely, Type 1 cephalocele, where the dural covering comprised of arachnoid tissue and few blood vessels limited to the stalk of the lesion and Type 2 cephalocele, where the meningeal tissue admixed with dermal and fibrous tissue and also few blood vessels were seen both in stalk as well as dome of the lesion. Patterson *et al.*[4] differentiated two groups based on the presence of vertical embryonic positioning of the straight sinus in six out of eight children. The features of vertical embryonic positioning of the straight sinus, "spinning-top" or peaking configuration of the tentorial incisura, CSF tract ("cigar"-shaped) within the interhemispheric fissure, and fenestration of the superior sagittal sinus in ACs were associated with poorer outcome. Brunelle *et al.*[5] reviewed 31 cases and found that 27 parietal ACs were all associated with persistent falcine sinus.

Morioka *et al.*[6] found CSF-tracts in three cases of ACs coursing through the superior sagittal sinus to the skull defect. The failure of mesodermal interposition between the dermal and neural ectoderm as the causative mechanism of AC and the persistent falcine sinus was considered as a marker of the timing of the embryologic insult that resulted in AC. In a review of 41 publications with 208 patients, Demir *et al.*[2] showed that the common locations are parietal (105/55%), occipital (71/37%), parieto-occipital (8), frontal (4), asterion (1), and sincipital (1). CNS anomalies were noted in 73.3% of parietal ACs and 32.4% of occipital ACs.

In a recent review of 30 reports with 68 parietal AC cases, Sencer *et al.*[7] found the following venous anomalies; fenestrated SSS in 48%, persistent falcine sinus in 47%, vertical embryonic positioning of the straight sinus in 44%, absent straight sinus in 39.7%, and anomalies in vein of Galen in 26.8%. Only 28 (38%) underwent surgical excision and the remaining 42 (62%) were followed-up conservatively. Majority of those followed up (20 of 24) had a good outcome.

In our series, three of them were occipital ACs, one each frontal and parietal AC. Three cases were associated with other CNS abnormalities. Only one parietal AC had

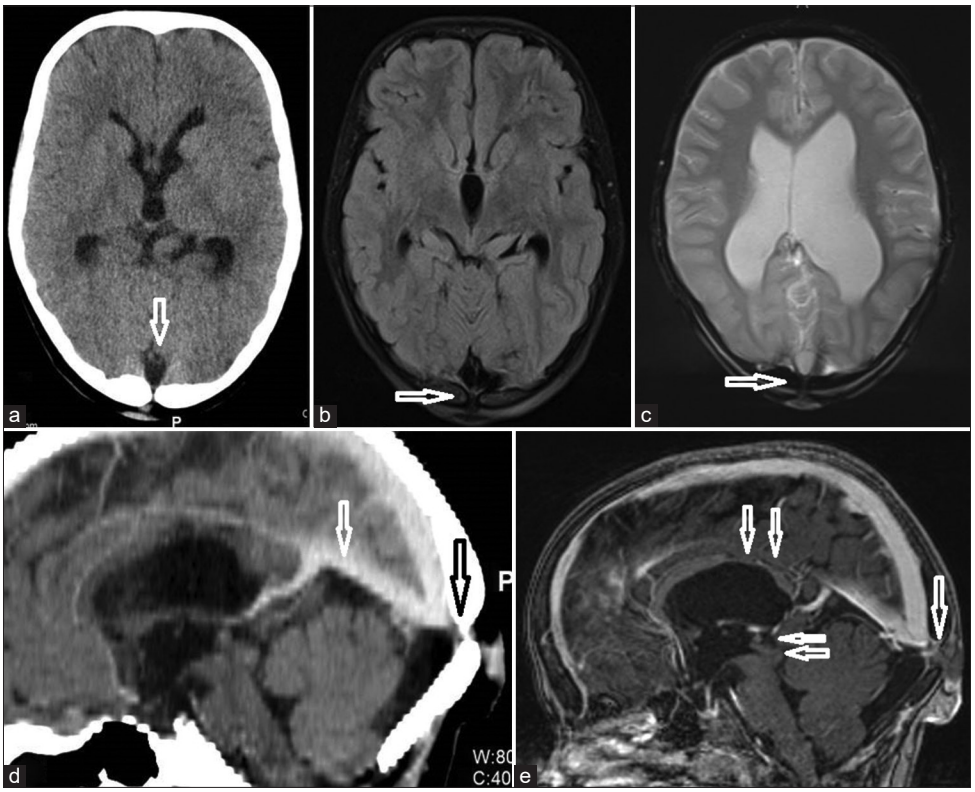

Figure 1: Case 1. Plain CT axial (a), MRI fluid attenuated inversion recovery axial (b), GRE T2* axial (c), Showing occipital bone defect at the level of Torcula. No herniation of CSF or neural tissue noted. Thin fibrous structure is seen extending from the meningeal margin into the defect. Contrast CT sagittal reformation (d), and contrast MRI T1-weighted images showing normal straight sinus along with hypoplasia of posterior corpus callosum and beaked tectal plate (vertical and horizontal double arrows in (e). MRI: Magnetic resonance imaging, CT: Computed tomography, CSF: Cerebrospinal fluid.

embryonic straight sinus. The differential diagnosis of ACs includes dermoid cyst, sinus pericranii, lipoma, and cephalhematoma. On computed tomography, the skull defect in dermoid is seen broader in outer table as compared to inner table, whereas in AC, it is vice versa. Sinus pericranii is an unusual communication between intracranial dural sinuses and extracranial venous structures through diploic veins. Surgery is indicated for cosmetic reasons or pain due to stretching of dura or overlying skin erosion or infection.

The ACs may have different clinical phenotypes based on location (parietal or occipital), associated intracranial neural or vascular anomalies, and are probably determined by environmental or dietary factors also apart from genetic predispositions.

CONCLUSION

The atretic cephaloceles may have different clinical phenotypes based on location (parietal or occipital), associated intracranial neural or vascular anomalies, and are probably determined by environmental or dietary factors also apart from genetic predispositions.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent.

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Conflicts of interest

There are no conflicts of interest.

REFERENCES

1. 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.
2. Demir MK, Çolak A, Ekşi MS, Özcan-Ekşi EE, Akakın A, Yılmaz B. Atretic cephaloceles: A comprehensive analysis of historical cohort. Childs Nerv Syst 2016;32:2327-37.
3. 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.
4. Patterson RJ, Egelhoff JC, Crone KR, Ball WS Jr. Atretic parietal

cephaloceles revisited: An enlarging clinical and imaging spectrum? AJNR Am J Neuroradiol 1998;19:791-5.
5. Brunelle F, Baraton J, Renier D, Renier D, Teillac D, Simon I, *et al*. Intracranial venous anomalies associated with atretic cephaloceles. Pediatr Radiol 2000;30:743-7.
6. Morioka T, Hashiguchi K, Samura K, Yoshida F, Miyagi Y, Yoshiura T, *et al*. Detailed anatomy of intracranial venous anomalies associated with atretic parietal cephaloceles revealed by high-resolution 3D-CISS and high-field T2-weighted reversed MR images. Childs Nerv Syst 2009;25:309-15.
7. Sencer S, Arnaout MM, Al-Jehani H, Alsubaihawi ZA, Al-Sharshahi ZF, Hoz SS. The spectrum of venous anomalies associated with atretic parietal cephaloceles: A literature review. Surg Neurol Int 2021;12:326.

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