The spectrum of venous anomalies associated with atretic parietal cephaloceles: A literature review

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INTRODUCTION

A “cephalocele” is a congenital herniation of intracranial contents through defects in the dura and cranium. There are four classes of cephaloceles, namely, meningoceles, meningoencephaloceles (encephaloceles), meningoencephalocystoceles (with parts of the ventricles), and atretic cephaloceles (ACs). ACs differ from true cephaloceles by the presence of dural remnants, fibrous tissue, as well as dysplastic neuronal tissue and are thought to represent involuted true cephaloceles (encephaloceles and meningoceles). ACs are rare lesions, accounting for 1% of all
cerebrospinal congenital anomalies and 37.5–50% of all cranial cephaloceles.\textsuperscript{37} ACs are further subdivided into “parietal” and “occipital” forms, in relation to their location adjacent to the vertex or the external occipital protuberance, respectively.\textsuperscript{37}

The concept of atretic parietal cephaloceles (APCs) as a separate entity was first investigated by Yokota et al.,\textsuperscript{37} who defined them as small, skin-covered, noncystic, nodular, or flat vertex midline lesions. Parietal atretic cephaloceles (PACs) are distinct in their frequent associating midline cerebral and venous malformations that govern their presentation, management, and prognosis.\textsuperscript{37} The description of these venous malformations becomes blurred when it comes to occipital cephaloceles, considering the higher degree of anatomical venous variations possible in their vicinity.

A wide range of APC-associated venous anomalies has been reported using the magnetic resonance venography (MRV) techniques. In this review, we intend to examine the available literature on the venous anomalies that may coexist with APCs. Our second objective is to report a new venous anomaly associated with an adult PAC presented in our exemplary case.

**MATERIALS AND METHODS**

The PubMed Medline database was searched using the following search algorithm: (((atretic cephalocele [Title/Abstract]) OR (Rudimentary meningocele, [Title/Abstract])) OR (Atypical meningocele [Title/Abstract]) OR (meningocele manqué [Title/Abstract]) OR (meningeal heterotopia [Title/Abstract])). No study type or time restrictions were applied. The initial search yielded a total of 85 articles. The abstract screening was used to identify articles reporting on APC. This was followed by a detailed review to include only papers describing the associated venous anomalies if present. Besides, the reference section of these papers was screened for relevant citations. Paper identification was done by two independent reviewers. Any discrepancy was resolved by discussion or the opinion of a third reviewer. Information extraction was based on the following parameters: study characteristics (author, year), patient age, concurrent venous anomalies, management approach (conservative vs. surgical), and patient outcomes [Table 1].

**RESULTS**

Our literature review yielded a total of 85 articles reporting on APC.\textsuperscript{[1,4,6,8,17,20,24,30,32,35–37,39]} Only those papers detailing the venous anomalies associated with APCs were included, for an overall of 30 articles [Table 1]. The total number of reported cases was 68 (including the exemplary case). The age range of the patients was 1 day–38 years, of whom 88% \((n = 62/68)\) belonged to the pediatric age group, and only 6 cases (12%) of adult PAC were reported. Two cases were diagnosed by second-trimester ultrasound.

The most commonly described associated venous anomaly was the presence of a “fenestrated superior sagittal sinus” (SSS) reported in 48.5% of the cases \((n = 33)\), followed closely by “persistent falcine sinus” (PFS) in 47% \((n = 32)\) and vertical embryonic positioning of the straight sinus (VEP SS) in 44% \((n = 30)\). The complete absence of the sagittal sinus (SS) was reported in 39.7% \((n = 27)\) and various abnormalities of the Galenic system were described in 26.8% of cases \((n = 12)\). Our exemplary case was the first to report an enlarged vein of Trolard.

Other reported venous malformations were partially absent SS and duplicated SSS, collectively reported in 4.4% of the cases \((n = 3)\).

Of all cases, only 38% underwent surgical excision \((n = 26)\), while the remaining 62% \((n = 42)\) were managed conservatively. Outcomes were reported for 35.3% \((n = 24)\) patients. Of those, the majority \((83\%, n = 20/24)\) had good outcomes, with “mild” neurological deficit, recurrence, and death occurring in 4.2%, 8.3%, and 4.2% \((n = 1/24, 2/24, and 1/24)\), respectively.

**Exemplary case**

A 25-year-old male presented to the outpatient clinic having a “scalp bulge.” On examination, the patient had a painless, small-sized midline mass situated in the parietal area. On palpation, the lesion was round, hard in texture, and fixed to surrounding structures, nontender, with intact overlying skin. The lesion was first noticed when the patient was 4 years old and has remained unchanged.

Brain computed tomography (CT) and magnetic resonance imaging (MRI) scans were ordered revealing a calvarium bifidum occupied by a heterogeneous cystic lesion with dysplastic brain parenchyma located in the frontal convexity.

![Figure 1](image_url)  
**Figure 1:** (a) Brain CT scan showing an enlarged left parietal cortical vessel, (b) sagittal brain CT scan showing a bulging at the vertex with protrusion of brain parenchyma inside and a cut in the SSS. CT.
## Table 1: The spectrum of APC-associated venous anomalies.

| Authors                      | Year   | Number of cases | Age range       | Associated venous anomalies                      | Management          | Outcome            |
|------------------------------|--------|-----------------|-----------------|-------------------------------------------------|---------------------|--------------------|
| Inoue et al.                  | 1938   | 3               | 6 m–7 y         | • Long vein of Galen (3)                         | Surgical excision   | Good               |
|                              |        |                 |                 | • VEP SS (3)                                     |                     |                    |
|                              |        |                 |                 | • VEP SS (6)                                     |                     |                    |
|                              |        |                 |                 | • Fenestration of SSS (4)                        |                     |                    |
| Patterson et al.              | 1998   | 8               | 1 d–3 y         |                                                 | Surgical excision   | 6 Normal           |
|                              |        |                 |                 |                                                 |                     | 1 with mild        |
|                              |        |                 |                 |                                                 |                     | motor delay        |
|                              |        |                 |                 |                                                 |                     | 1 died at 3 y      |
|                              |        |                 |                 |                                                 |                     | N/A                |
| Martínez-Lage et al.          | 1997   | 1               | 38 y            | VEP SS                                          | Conservative        |                    |
| McLone and De Leon[24]        | 1998   | 1               | 7 m             | Abnormal Galenic system                         | N/A                 | N/A                |
| Brunelle et al.[4]           | 2000   | 27              | N/A             | • Persistent falcine sinus (PFS) (27)           | N/A                 | N/A                |
|                              |        |                 |                 | • Absent straight sinus (23)                    |                     |                    |
|                              |        |                 |                 | • Fenestrated SSS (23)                          |                     |                    |
|                              |        |                 |                 | • Absent vein of Galen (4)                      |                     |                    |
| Morioka et al.[25]           | 2009   | 3               | 1–18 y          | VEP SS                                          | Surgical excision   | N/A                |
|                              |        |                 |                 |                                                 |                     |                    |
|                                 |        |                 |                 |                                                 | Surgical excision   | N/A                |
| Yoshida et al.[30]           | 2006   | 1               | 13 y            | VEP SS                                          | Surgical excision   | N/A                |
| Kim et al.[35]               | 2006   | 1               | 11 y            | VEP SS                                          | Surgical excision   | Good               |
|                              |        |                 |                 | • PFS                                           |                     |                    |
|                              |        |                 |                 | • Abnormal Galenic system                       |                     |                    |
|                              |        |                 |                 | • Absent straight sinus                         |                     |                    |
| Wang et al.[39]              | 2010   | 1               | 6 y             | VEP SS                                          | Surgical excision   | Good               |
| van Laak et al.[34]          | 2010   | 1               | 4 m             | VEP SS                                          | Conservative        | N/A                |
| Kumar et al.[34]             | 2010   | 1               | 4 y             | VEP SS                                          | N/A                 | N/A                |
| Şengöz et al.[30]            | 2011   | 1               | 23 y            | • VEP SS                                        | Conservative        | N/A                |
|                              |        |                 |                 | • Abnormal Galenic system                       |                     |                    |
|                              |        |                 |                 | • Abnormal straight sinus                       |                     |                    |
|                              |        |                 |                 | • PFS                                           |                     |                    |
| Hsu et al.[12]               | 2011   | 1               | 16 y            | VEP SS                                          | Surgical excision   | N/A                |
|                              |        |                 |                 |                                                 |                     |                    |
|                                 |        |                 |                 |                                                 | Surgical excision   | N/A                |
| Yilmaz et al.[36]            | 2011   | 1               | 36 y            | VEP SS                                          | Surgical excision   | Recurrence         |
| Muralidharan et al.[27]      | 2013   | 1               | 22 m            | VEP SS                                          | Surgical excision   | N/A                |
|                                 |        |                 |                 |                                                 |                     |                    |
|                                 |        |                 |                 |                                                 | Surgical excision   | N/A                |
| Leykamm et al.[17]           | 2013   | 1               | 1 d             | VEP SS                                          | Surgical excision   | Good               |
| Siverino et al.[31]          | 2015   | 1               | 3 y             | VEP SS                                          | Surgical excision   | Good               |
|                              |        |                 |                 | • Fenestrated SSS                                |                     |                    |
|                              |        |                 |                 | • Fenestrated SSS                                |                     |                    |
|                              |        |                 |                 | • Abnormal Galenic system                       |                     |                    |
| Bick et al.[3]               | 2015   | 1               | 6 m             | VEP SS                                          | Surgical excision   | N/A                |
| Anand et al.[1]              | 2015   | 1               | 4 w             | VEP SS                                          | Surgical excision   | Good               |
| Ertuğrul et al.[8]           | 2015   | 1               | 25 y            | VEP SS                                          | Surgical excision   | Recurrence         |
| Kumar et al.[14]             | 2016   | 1               | 1 y             | VEP SS                                          | N/A                 | N/A                |
|                              |        |                 |                 | • Fenestrated SSS                                |                     |                    |
|                              |        |                 |                 | • Fenestrated SSS                                |                     |                    |
|                              |        |                 |                 | • Abnormal Galenic system                       |                     |                    |
| Gulati et al.[11]            | 2016   | 1               | 6 m             | VEP SS                                          | Surgical excision   | Good               |
|                              |        |                 |                 | • Fenestrated SSS                                |                     |                    |
|                              |        |                 |                 | • Fenestrated SSS                                |                     |                    |
| Santos et al.[29]            | 2016   | 1               | 22 w (intrauterine) | VEP SS                                        | Surgical excision   | Good               |
| Demir et al.[6]              | 2016   | 1               | 47 y            | • Fenestrated SSS                                | Conservative        | N/A                |
| Murakami et al.[26]          | 2017   | 1               | 1 m             | • VEP SS                                        | Surgical excision   | Good               |
|                                 |        |                 |                 | • SS and SSS deviated to right                  |                     |                    |
|                                 |        |                 |                 | • VEP SS                                        |                     |                    |
| Gagliardo et al.[18]         | 2018   | 1               | Newborn         | • Duplicated SSS                                 | Conservative        | Good               |
| Siverino et al.[31]          | 2015   | 1               | 23 w (prenatal) | VEP SS                                          | Surgical excision   | Good               |
|                              |        |                 |                 | • Fenestrated SSS                                |                     |                    |
|                              |        |                 |                 | • Fenestrated SSS                                |                     |                    |
|                              |        |                 |                 | • Enlarged vein of Trolard                       |                     |                    |
| Tsitouridis et al.[32]       | 2005   | 2               | 2 m             | PFS                                             | N/A                 | N/A                |
|                              |        |                 | 8 m             |                                                 |                     |                    |
| Favoroel et al.[9]           | 2015   | 1               | 1 y             | • Absent SS                                      | Conservative        | N/A                |
| Present case                 | 2020   | 1               | 25 y            | • Abnormal Galenic system                       | Conservative        | Good               |
|                              |        |                 |                 | • Fenestrated SSS                                |                     |                    |
|                              |        |                 |                 | • Enlarged vein of Trolard                       |                     |                    |

**Notes:**

- VEP SS: Vertical embryonic positioning of the straight sinus
- SS: Straight sinus
- SSS: Superior sagittal sinus
- PFS: Persistent falcine sinus
- APC: Atretic parietal cephalocele
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The MRV revealed an anomalous vein, along with a focal indentation of the SSS on the left side, suggesting a vascular malformation. Next, a CT angiogram (CTA) was done, revealing a large oval filling defect in the middle third of the SSS (about 44 × 22 mm) with some opacified intrinsic filling vessels. The CTA also showed a grossly dilated left cortical parietal vein extending from the Sylvian fissure to open in the SSS at the site of the filling defect [Figure 3]. At this point, type 1 dural arteriovenous fistula (DAVF) and a PAC with associated venous anomalies were the two main differentials.

A digital subtraction angiography was performed which excluded DAVF and the diagnosis of an enlarged vein of Trolard associated with fenestration of the SSS in the setting of a small atretic parietal encephalocele was made [Figure 4]. The patient was managed conservatively. His follow-up MRI scans at 6 months and 2 years showed no new findings.

DISCUSSION

Nomenclature and classification of ACs

Alternative terms that have been used to describe ACs include meningocele manqué, abortive, rudimentary, occult, sequestered, atypical meningocele, and atypical heterotopia. Although these terms have been used interchangeably, differences between them do exist, as identified by Lopez et al. [18]

Martinez-Lage et al. classified ACs into two types. Type 1, which is limited to the stalk of the lesion, contains arachnoid tissue as well as tangles of anomalous blood vessels and is covered by hairy skin. Type 2 extends to the dome of the lesion and consists of meningeal tissue intermingled with dermal and fibrous tissue, as well as clusters of anomalous blood vessels, extending as a net, and ectopic neural or glial element. [20-22] For the majority of cases, no pathological data were available and this parameter was hence excluded from the final analysis.
Evolving terminology for atretic parietal cephalocele (APC)-associated venous anomalies

The cooccurrence of APCs with venous malformations was first described by McLaurin et al. in 1964 who described anomalies such as fenestrated SSS and VEP SS.[23]

Typical angiographic features of VEP SS include elongated internal cerebral veins that join a small single vein the cistern (vein of Galen), which coalesces with a shortened vertically positioned SS in the faix to join the SSS.[26] Before the advent of CISS and 3T-T2R MRI sequences, the detailed course of these anomalies was less clearly delineated. For example, the elongated ICVs were referred to as an “elongated vein of Galen.” In other studies, conventional MRI images failed to show the SS, and thus, “absent SS with an anomalous Galenic system” was the reported anomaly.

Etiopathogenetic and embryonic correlations of APCs and their associated venous anomalies

Both the etiology and embryological basis of APC and their associated venous anomalies are currently a source of controversy. Both genetic and environmental factors are thought to play a role in the pathogenesis of AC, including Vitamin A, teratogens, X-ray, folic acid antagonists, trypan blue, triamcinolone, and malnutrition.[20-22] Familial cases of ACs have also been rarely reported, although an exact genetic marker has not been identified.[5,19,39]

As to their embryonic origin of the APCs, several theories have been proposed, including failure of neural tube closure, normal closure followed by an abnormal reopening of the neural tube, partial regression of an intrauterine meningoencephalocele, a sequel of a primary mesenchymal injury, the persistence of neural crest remnants, or a nuchal bleb.[3,5,7,21,33] Figure 5. The presence of both skin and neural defects suggests a shared embryonic origin for the malformations since both tissues are derived from the ectoderm.

Figure 4: (a and b) Digital (a) and native (b) images of the cerebral angiogram show fenestration of the superior sagittal sinus due to interposition of the encephalocele also causing a bony defect in the convexity with enlarged vein of Trolard on the left side.

Figure 5: The embryologic pattern of the intracranial venous system.
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Fenestration of the SSS was the most commonly reported APC-associated venous anomaly. The SSS forms during the 35–50 gestational days from the fusion of the marginal sinuses. The presence of the AC and its associated fibrous tracts is thought to interrupt this process, resulting in splitting or “fenestration” of the SSS.[31]

**Surgical perspective**

APCs typically present as a skin-covered midline scalp mass.[35] Clinically, most patients with atretic encephaloceles are asymptomatic and usually discovered incidentally. Surgery is advised for esthetically unpleasant lesions, those with, or at high risk of, ulceration or rupture due to their prominent location, masses causing headache due to stretching of the dura, and lesions requiring histological diagnosis.[35]

Preoperative imaging of both the superficial and deep venous systems is mandatory in cases of APCs, given the high incidence of associated venous anomalies. Interestingly, while these venous anomalies rarely necessitate formal intradural exploration in pediatric practice, intradural extension was required in our case, raising the question of whether these anomalies progress over time and, thus, the importance of surgery timing. The surgery typically involves excising of the mass with cranioplasty, without intervening with the venous anomalies, as they participate in normal venous drainage. In the absence of other intracranial anomalies, the prognosis of APCs is generally good.[31]

**Study limitations and future research directions**

One limitation of this review is that the majority of the studies were single case reports or small series, with limited data on patient outcomes. For a robust conclusion on the prognostic significance of specific venous anomalies, larger multicenter, outcome-oriented studies are required. Furthermore, more angiographic studies that could better delineate the anatomy of the associated venous anomalies and address the inconsistencies in the terminology used to describe them are the way forward toward a better understanding of APCs.

**CONCLUSION**

Although benign in nature, PACs are also a marker for the presence of a spectrum of cerebral venous malformations of which the neurosurgeons and neuroradiologists should be vigilant. When PAC is suspected, a comprehensive preoperative imaging workup for both the superficial and deep venous systems is, therefore, required to obtain an accurate understanding of the course of the venous system and use these data to inform surgical planning.

**Declaration of patient consent**

Patient’s consent not required as patients identity is not disclosed or compromised.

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**Conflicts of interest**

There are no conflicts of interest.

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