Frontal intradiploic encephalocele in a 44-year-old male patient: illustrative case

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BACKGROUND Encephaloceles are protrusions of the cerebral tissue through a skull defect. They occur mostly in children and very rarely in adults.

OBSERVATIONS The authors present a case of a 44-year-old man presenting with a first-time generalized seizure. Computed tomography of the head showed bone destruction associated with a right frontal lesion. Magnetic resonance imaging scans demonstrated a largely isointense lesion in the intradiploic space that contained small, hyperintense nodular components and showed a low to moderate contrast agent enhancement.

LESSONS The patient underwent resection, during which the histological examination found the lesion to be an intradiploic encephalocele. The patient had an uneventful postoperative course with a cessation of seizures. The imaging and neuropathological findings as well as a literature review, together with a discussion on the etiology of intradiploic encephaloceles, are contained in this report.

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KEYWORDS encephalocele; adult; head trauma; epilepsy

Encephaloceles are defined as cephalic herniations through a defect of the dura mater or skull. Encephaloceles occur rarely in adults; they are more commonly found in children.1 The protrusion typically contains meninges, cerebrospinal fluid (CSF), and cerebral tissue.2,3 Encephaloceles are mostly congenital in origin (primary encephalocele) and develop due to failure of neural tube closure, eventuating in a bone defect through which the intracranial contents protrude. As congenital lesions, primary encephaloceles are generally present at birth or occur during childhood. They may also be acquired subsequent to traumatic, neoplastic, infectious, and metabolic damage as well as surgical procedures (secondary encephalocele).4,5

Primary encephaloceles generally occur in the midline. The occipital region is the most common location of these lesions.1,6 It has been reported that nearly 75% of primary encephaloceles are located in the occipital region, whereas only 13%–15% occur in the frontoethmoidal region and 10%–12% occur in the parietal or sphenoidal region.7,8 Patients usually exhibit direct neurological symptoms such as motor or sensory deficits parallelizing the involved cortical area of the traction or herniation. Seizures, recurrent meningitis, and CSF otorrhea are the other possible clinical presentations of this entity.8–10

As a congenital malformation, encephaloceles are very rare, with a prevalence of 0.8 to 5 per 10,000 live births; thus, they are less common than other neural tube defects.11 The actual incidence of acquired encephaloceles remains unknown. Nevertheless, it is assumed that the majority of encephaloceles are congenital and manifest at early ages.12 It is not obligatory for the encephalocele hernia to traverse the whole thickness of the skull. In some cases, the outer skull table remains intact while the cerebral herniation pierces the defective dura mater and the inner skull table into the intradiploic space. These cases are termed “intradiploic encephaloceles.”13 Sometimes, intradiploic encephaloceles show imaging findings similar to those of osteolytic lesions, leading to a wide variety of differential diagnoses, such as eosinophilic granuloma, plasmacytoma, osteosarcoma, cavernous hemangioma, dermoid cysts, or metastasis. The absence of overt malignancy features, the preserved integrity of the outer skull table, and the presence of leptomeninges in the lesion may imply benign cystic lesions such as intraosseous leptomeningeal cysts or intradiploic arachnoid cysts.14 However, none of
FIG. 1. Axial bone window CT scan (A) displays the defective inner table and the widened diploic space in the right frontal skull. Furthermore, a short segmentally thinned outer table can be seen.
these lesions enclose the cerebral parenchyma, which is characteristic of the intradiploic encephalocele. Here, we describe a case of a right frontal intradiploic encephalocele in a 44-year-old man with localized meningothelial proliferation reminiscent of meningioma and trauma history in the same region. The computed tomography (CT) and magnetic resonance imaging (MRI) findings and the histological examination exemplify the hallmarks of this rare type of lesion.

Illustrative Case

A 44-year-old, male, hitherto healthy patient presented to the emergency department after a first-time generalized seizure without any history or clinical signs of sleep deprivation or alcohol or drug consumption. The patient could not recall the convulsive activity in a subset of cortical neurons with slightly aberrant morphology (Fig. 1G-H). The ini- tiation of small reactive meningothelial proliferations. Due to the small amounts of meningothelial tissue and bland cytology, edges of the defect and lacked noticeably elevated mitotic frequency. Considering the clinical presentation as well as radiological and histological findings concerning this intradiploic lesion with osteolysis, the neuropathological diagnosis of a frontal intradiploic encephalocele was made.

Discussion

The term “intradiploic encephalocele” refers to the herniation of cerebral tissue through an inner table defect into the intradiploic space.13

Observations

With only 21 reported cases in the literature, including ours (Table 1), this is a rare type of pathology. Intradiploic encephaloceles exhibit features that clearly distinguish them from the more frequent cranium bifidum cases, that is, primary encephaloceles as a form of dysraphic lesions. Seventy-five percent of primary encephaloceles occur in the occipital region.7,8 By contrast, 12 (57.14%) of 21 intradiploic encephalocele cases were located parietally, 5 (23.81%) were observed in the occipital region, and 4 patients (19.05%) exhibited frontal intradi- ploic encephaloceles. Primary encephaloceles were reported to occur mostly in children. Conversely, the average age at diagnosis of intradiploic encephaloceles was 48.29 years, with only 4 of the 21 reported cases having been younger than 15 years old at the time of diagnosis (Table 1). In a synopsis of the reported intradiploic ence- phalocele cases, we can maintain that intradiploic encephaloceles arise predominantly in adults and in the parietal region, which clearly differs from primary encephaloceles, which occur at a young age and mostly in the occipital region.

The histological and immunohistochemical examinations revealed brain tissue with gray and white matter exhibiting chronic reactive tissue alterations, including marked fibrillary astrogliosis and the presence of foamy macrophages. Furthermore, the blood vessels in the herniated brain tissue showed reactive alterations in the form of moderate vessel wall fibrosis. Hemosiderin deposits as a sign of chronic blood–brain barrier alterations were found in the herniated central nervous system (CNS) parenchyma, mainly in perivascular macrophages. The CNS tissue was surrounded by bone. Furthermore, fragments of the dura mater with small, finger-like infiltrations of meningothelial proliferations adhering to intradiploic brain tissue were apparent (Fig. 1G-L). The meningothelial proliferations were most prominent at the bony edges of the defect and lacked noticeably elevated mitotic frequency. Due to the small amounts of meningothelial tissue and bland cytology, the diagnosis of a meningioma was dismissed in favor of the interpretation of small reactive meningothelial proliferations.

FIG. 1. Axial brain window CT scan (B) shows a radiodensity comparable to brain parenchyma within the intradiploic lesion. Sagittal T1-weighted image (C) shows a lesion in the calvarial vault, which is widely hypointense and partly isointense compared with CNS parenchyma and exhibits low to moderate contrast enhancement (D). The lesion appears mostly isointense with the brain cortex on the T2-weighted dark fluid sequence (E). It also contains a small nodular hyperintense component. F: The diffusion-weighted imaging sequence (apparent diffusion coefficient mapping) displays a CSF-filled diploic space as well as a small nodular component with an intensity of brain parenchyma. Histological and immunohistochemical examination of the resected lesion reveals brain tissue with perivascular hemosiderin deposits and surrounded by bone (G and J). Finger-like infiltrates of meningothelial meningioma adhering to intradiploic brain tissue and staining for epithelial membrane antigen (EMA) were found (G and H). Immunohistochemistry for myelin basic protein (MBP) displays largely normal gray and white matter myelin structure within the encephalocele (K). However, the SMI-31 staining reveals moderate positivity in a subset of cortical neurons with slightly aberrant morphology (L), indicating chronic neuronal damage in the herniated brain tissue. Original magnification of histological images: ×10 (G), ×10 (H), ×20 (inset, H), ×20 (I), ×10 (J), ×20 (inset, J), ×10 (K), ×10 (L), and ×20 (inset, L). Scale bars = 1 mm (G, H, J, K, and L) or 500 μm (insets and I). H&E = hematoxylin and eosin.
| Authors & Year          | Age, Sex | Location                  | Signs & Symptoms                          | Examination of Skull | Imaging       | Hx of Trauma | Surgery                                                                 | Follow-Up                      |
|------------------------|----------|---------------------------|-------------------------------------------|----------------------|---------------|--------------|--------------------------------------------------------------------------|--------------------------------|
| Kosnik et al., 1976    | 57 yrs, M| Parietal, lt              | Generalized tonic-clonic Sz, expressive aphasia | Normal               | Skull radiograph | No           | Yes, excision of cerebral herniation                                  | Asymptomatic                   |
| Patil et al., 1996     | 64 yrs, M| Parietal, lt              | Increasing lump on his head                | Lump                 | CT & MRI      | Yes, 1 yr prior to admission | Yes, decompressive surgery                                         | Asymptomatic                   |
| Martinez-Lage et al., 1997 | 6 yrs, F | Frontal, rt              | Tingling & lt hemiparesis, rt frontal pain | Lump                 | Skull radiograph, CT, & MRI | Iatrogenic/ craniostenosis surgery | Yes, excision of cerebral herniation                                  | Asymptomatic                   |
| Lenthall et al., 1999  | 15 mos, NR| Occipitoparietal, rt     | Pulsatile swelling over occipital region   | Lump                 | Skull radiograph & MRI | Yes, 9 mos prior to admission | Yes, NR NR                                                            | NR                              |
| Peters et al., 2002    | 36 yrs, M| Parietal, lt              | Problems of rt leg coordination            | Normal               | MRI & fmMRI   | No           | Yes, excision of cerebral herniation                                  | Asymptomatic                   |
| Aterrietehau et al., 2004 | 73 yrs, F | Parietal, rt              | HA                                         | NR                   | CT & MRI      | No           | No NR                                                                  | NR                              |
| Froelich et al., 2006  | 51 yrs, F| Parietal, rt              | Hemiparesthesia, lt                       | Lump                 | Skull radiograph, CT, & MRI | No           | Yes, excision of cerebral herniation                                  | Symptomatic/persistent hemiparesthesia, lt |
| Tsuboi et al., 2006    | 66 yrs, M| Parietal, rt              | Dizziness                                 | Normal               | CT, MRI, & SPECT | No           | No                                                                     | Asymptomatic                   |
| Loumiotis, et al., 2010| 50 yrs, M| Parietal, lt              | Rt arm weakness                           | NR                   | CT & MRI      | No trauma Hx, but violent coughing spell, Yes, decompressive surgery | Progressive yet incomplete clinical improvement | |
| Dobrin et al., 2011    | 75 yrs, M| Parietal, lt              | Partial Szs in rt lower limb              | Normal               | CT & MRI      | No           | Yes, excision of cerebral herniation                                  | Asymptomatic                   |
| Kim et al., 2015       | 50 yrs, M| Parietal, lt              | HA                                        | Normal               | Skull radiograph, CT, MRI, & angiography | No           | No                                                                     | Asymptomatic                   |
| Arevalo-Perez et al., 2015 | 84 yrs, F | Parietal, rt              | Progressive disorientation, nonspecific   | NR                   | CT & MRI      | No           | No                                                                     | NR                              |
| McPheeters et al., 2015| 60 yrs, F| Frontal, rt               | Generalized tonic-clonic Sz                | Lump                 | CT & MRI      | Yes, multiple times as young adult | Yes, excision of cerebral herniation                                | NR                              |

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TABLE 1. Systematic overview of reported intradiploic encephalocele cases since 1976

| Authors & Year          | Age, Sex | Location | Signs & Symptoms | Examination of Skull | Imaging | Hx of Trauma | Surgery | Follow-Up          |
|-------------------------|----------|----------|------------------|----------------------|---------|-------------|---------|-------------------|
| Mazzucchi et al., 2016  | 38 yrs, M| Parietal, lt | HA, vomiting & hypoesthesia of rt arm | Normal | CT & MRI | Yes, 23 yrs prior to admission | Yes, excision of cerebral herniation | Asymptomatic |
| Shi et al., 2017        | 45 yrs, M| Parietal, rt | Hemiparesis lt | Normal | CT & MRI | Yes, 36 yrs prior to admission | Yes, w/o excision | Progressive yet incomplete clinical improvement |
| Valci et al., 2018      | 70 yrs, M| Frontal, rt | Spastic progressive paresis of lt lower limb | NR | CT, MRI, fMRI, & SPECT | NR | Yes, decompressive surgery | Asymptomatic |
| Vavro et al., 2018      | 77 yrs, F| Occipital, rt | Dizziness & unsteadiness | NR | CT & MRI | NR | NR | NR |
| Chakkalakkoombil et al., 2018 | 52 yrs, F| Occipital, rt | HA | Normal | MRI | Yes, 1 yr prior to admission | NR | NR |
| Kandemirli et al., 2019 | 11 yrs, M| Occipital, lt | Generalized tonic-clonic Szs | NR | CT & MRI | Yes, 8 yrs prior to admission | No | NR |
| Chen et al., 2021       | 8 yrs, M | Occipital, lt | 2 episodes of loss of consciousness | Normal | CT & MRI | Yes, 7 yrs prior to admission | No | Asymptomatic |
| Present case            | 44 yrs, M| Frontal, rt | Generalized tonic-clonic Szs | Normal | CT & MRI | Yes, 5 yrs prior to admission | Yes, excision of cerebral herniation | Asymptomatic |

HA = headache; Hx = history; NR = not reported; SPECT = single-photon emission computed tomography; Sz = seizure.
Cases in this table are only those listed in PubMed under the exact diagnosis of “intradiploic encephalocele” and “intradiploic meningoencephalocele” since 1976. Similar case descriptions with different nomenclature were not included.
The frequent presentation at adult age, frequent atypical locations aside from the brain midline, and the lack of accompanying malformations render a congenital cause of intradiploic encephaloceles unlikely. A very plausible concept by Patil et al. postulates blunt head injuries (as in our case) with low velocity as the origin of inner table breakage, because the inner table is thinner than the outer table. Inwardly directed fractured bony edges further lead to the tearing of the dura mater and arachnoid mater. The recolling of the fractured edges subsequently leads to negative pressure, drawing brain tissue and CSF into the intradiploic space. After all, physiologically pulsating CSF might explain slow expansion of the intradiploic space. This concept seems all the more plausible because, in our case, we observed a liquid-filled intradiploic cyst that was considerably larger than the herniated brain tissue. A history of head trauma was documented in 9 (including our case) of 21 reported cases in the literature (Table 1). It is possible that a head trauma might not always have been experienced as significantly painful and might not be remembered, especially if it occurred many years prior. Therefore, the absence of a trauma history does not preclude the possibility of an intradiploic encephalocele. Indeed, Peters et al. assumed that the intradiploic encephalocele present in their patient was caused by trauma, even though there was no definite trauma history.

Another characteristic in our case is small fragments of meningothelial proliferations at the margins of the bony tissue adhering to the intradiploic cerebral tissue (Fig. 1). In the literature, Valci et al. described a meningioma accompanying an intradiploic encephalocele. Also in that case, the meningothelial proliferations adhered to the margins of the bony fragments encountered around the encephalocele. Valci et al. described this meningioma as World Health Organization grade 1. High mitotic activity was thus detected neither there nor in our present case. This raises the possibility that the meningioma reported by Valci et al. was also not an actual neoplasm but represents reactive arachnoidal proliferations induced by chronic irritation of the arachnothelial membrane along the fractured edges of the inner table, as we have described above.

However, if we assume a nonneoplastic origin, and rather a proliferation of the arachnoid membrane due to long-term irritation by the fractured edges of the inner table, patients with intradiploic encephalocele should more commonly present with meningothelial proliferations than only the 2 reported cases (including our case). Still, we must take into account the rarity of this entity as well as the fact that not every patient underwent surgical intervention or a fully reported and extensive histological examination.

In most cases, surgery is the method of choice for both definite diagnosis and therapy. Commensurately, 13 of 21 patients (including our case) are reported to have undergone neurosurgery including parenchymal excision and decompressive surgery. Ten of these operated patients achieved a sustained release of symptoms, whereas 3 of them (2 decompression surgery and 1 excision surgery) remained symptomatic. Peters et al. and Valci et al. performed functional MRI (fMRI) examinations showing lack of task-related activation within the encephalocele; however, activation was found within the neighboring regions. Thus, they concluded this symptoms were probably caused not by the encephalocele itself but through the progressive stretching and elongation of the brain parenchyma in the diploe.

Lessons
Tsdboi et al. conducted a single-photon emission computed tomography examination of the encephalocele tissue that showed the same signal pattern as normal brain tissue. Our case displayed a tissue structure that was partly comparable to normal brain architecture (Fig. 1, myelin basic protein). Furthermore, Kim et al. in addition to CT and MRI, also performed cerebral angiography, which exhibited a normal cerebral vessel pattern in the encephalocele. In the synopsis of the previous cases and our examinations, intradiploic encephaloceles can be assumed to exhibit largely normal cerebral vascular structure as well as brain tissue with reactive changes such as gliosis as well as chronic neuronal injury. Here, the reported fMRI results deserve further consideration. Lack of detectable motor-related activation in the encephalocele itself does not necessarily reflect non-functionality of the herniated brain parenchyma but may rather indicate an altered blood flow response in the calvarial vault. Correspondingly, our histology and further radiological examinations as well as the results of cortical stimulation of Valci et al. exhibited largely normal cerebral tissue.

The treatment of patients manifesting without neurological deficits remains controversial. If a patient remains asymptomatic or responds to symptomatic therapy, observation and regular follow-up seem appropriate.

Intradiploic encephaloceles are an extremely rare type of lesion with characteristics distinguishing them clearly from the classic encephaloceles. Our case report adds to the literature with some unusual features and presents a systematic review of the literature that includes all reported cases since 1976.

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Author Contributions
Conception and design: Hainfellner, Atli, Hametner. Acquisition of data: Hainfellner, Atli, Rath, Hametner. Analysis and interpretation of data: Atli, Hametner. Drafting the article: Atli, Hametner. Critically revising the article: Hainfellner, Atli, Rath, Burtscher, Hametner. Reviewed submitted version of manuscript: Atli, Rath, Hametner. Administrative/technical/material support: Atli, Burtscher. 

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