■ Case vignette

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Epileptic Disorders

Encephalocele-associated temporal lobe epilepsy and skull fibrous dysplasia: a report of two cases

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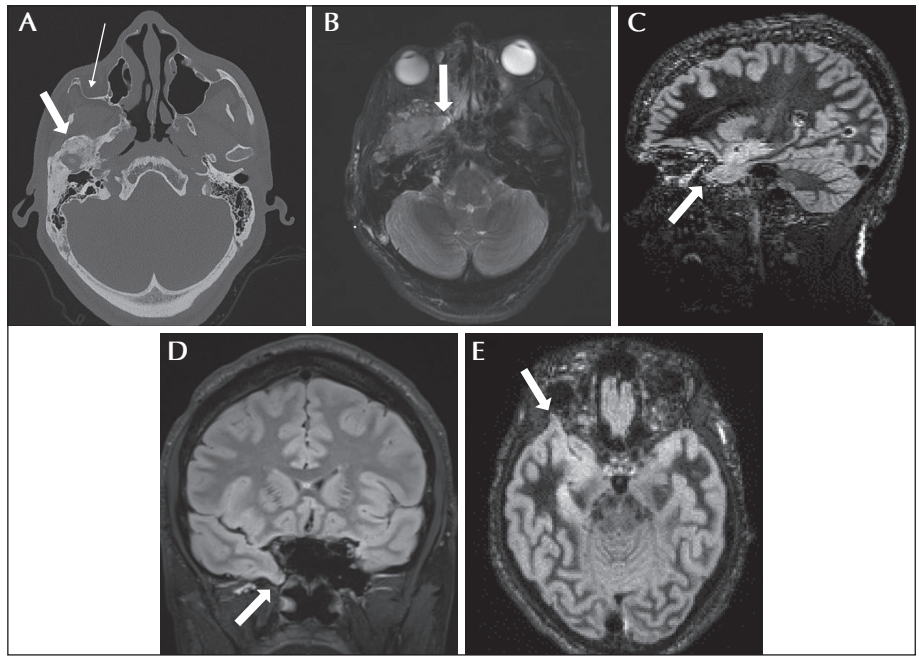
Temporal lobe encephaloceles are an important cause of drug-resistant focal epilepsy, and advances in imaging modalities have led to increased recognition [1, 2]. Fibrous dysplasia (FD) is a benign process in which bone is replaced by connective tissue and poorly formed trabecular bone. It may affect multiple bones (polyostotic) or remain isolated to one bone, including the skull [3]. Although seizures have been recognized in patients with FD of the skull [4-9], the pathogenicity has not been well understood. Here, we describe two cases of temporal lobe epilepsy in patients with FD of the skull with associated encephaloceles that were successfully treated surgically.

A 19-year-old man with polyostotic FD presented with drug-resistant focal epilepsy with seizure onset at age 10. FD was diagnosed in childhood due to an asymmetric jaw and failure of eruption of dentition. He had focal seizures which involved dysgeusia, spitting, visual blurring and difficulty focusing with occasional focal to bilateral tonic-clonic seizures (FBTCS). Seizures continued despite treatment with multiple antiseizure medications (ASMs). CT revealed the known polyostotic FD of the right petrous and squamosal temporal bone and sphenoid wing. MRI demonstrated an associated right temporal, sphenoid wing encephalocele adjacent to the FD (*figure 1*). Right temporal sharp waves and one right temporal seizure were recorded on EEG. The patient underwent an inferior temporal resection and reconstruction of the middle fossa floor. Multiple encephaloceles were also noted intraoperatively that were retrospectively identified on imaging, and these were reduced during surgery. Seizures persisted after the initial surgery. Therefore, he underwent right anterior temporal lobectomy and amygdalohippocampectomy (ATLAH) which resulted in seizure freedom at one-year of follow-up.

A 30-year-old man presented with seizure onset at age 21 with a FBTCS. He then developed recurrent focal seizures with impaired awareness, speech arrest, and oral automatisms, and continued to have rare nocturnal FBTCS, which persisted despite trials of multiple ASMs. MRI demonstrated a large right temporal, sphenoid wing encephalocele (*figure 2*). FD of the right temporal bone and sphenoid wing was confirmed on skull base CT, producing a skull defect containing the encephalocele. EEG monitoring revealed right temporal sharp waves and three right temporal seizures (*supplementary figure 1*). The patient underwent encephalocele repair and temporal tip resection which has resulted in seizure freedom for six weeks according to the last follow-up visit.

We describe two cases of FD of the skull base with associated encephaloceles and resultant drug-resistant epilepsy. Both cases had seizure onset ipsilateral to the encephaloceles and were



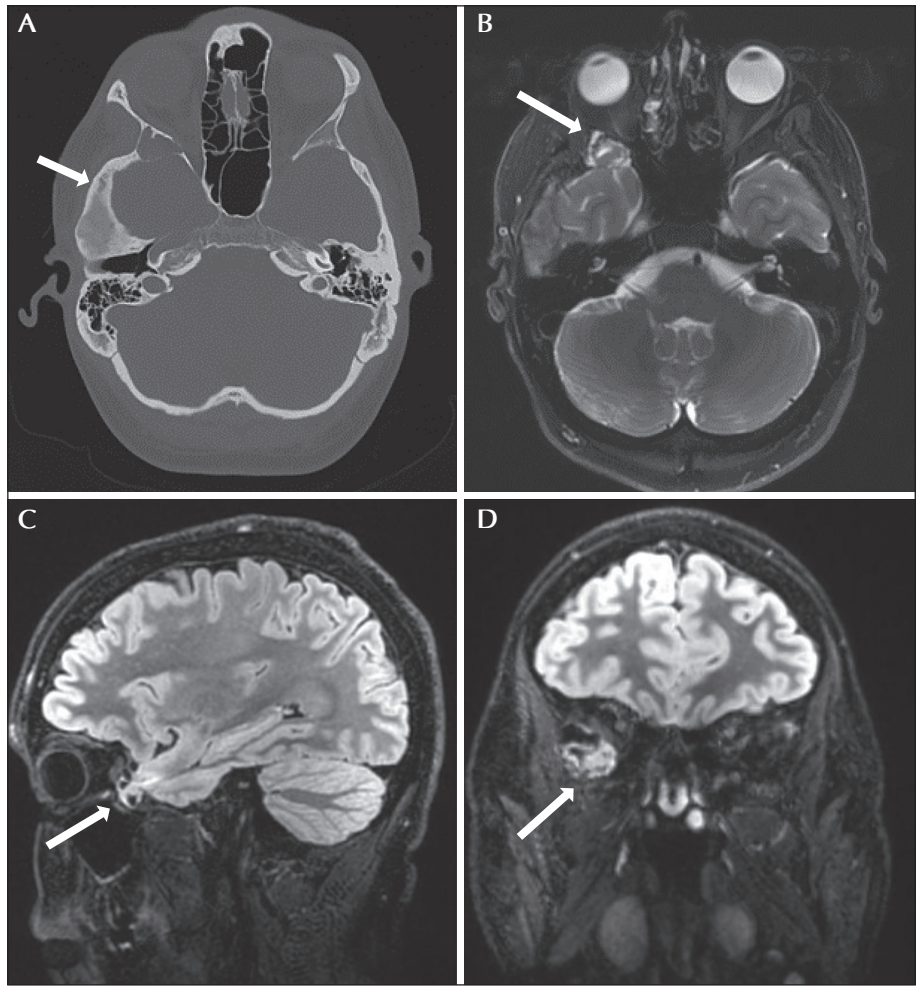
■ **Figure 1.** Fibrous dysplasia with encephaloceles in Case 1. (A) Head CT demonstrates mottled ground-glass expansion of the right petrous temporal bone (arrow) and a dysplastic right sphenoid wing (thin arrow), consistent with fibrous dysplasia. (B-D) MRI (axial, sagittal, and coronal sequences) shows a right sphenoid wing encephalocele (arrow) with cerebral parenchyma herniating inferiorly-medially into an osseous defect. (E) MRI (axial) shows an additional, more lateral, temporal encephalocele (arrow). There were also changes of fibrous dysplasia in the right mandible (not shown).

successfully treated surgically, supporting that the encephaloceles were causative of seizures. However, Case 2 only had short-term follow-up information available limiting the conclusion about outcome. In addition, an association between FD or encephaloceles and other subtle central nervous system abnormalities leading to seizures is a possibility that cannot be completely excluded. To our knowledge, the association between encephalocele formation and FD, leading to drug-resistant epilepsy, has not been previously described.

FD is a developmental anomaly of the bone in which normal bone is replaced by softer fibrous tissue [3]. FD is caused by a postzygotic mutation in the guanine nucleotide stimulatory protein gene leading to increased cyclic adenosine monophosphate (cAMP) and arrest of differentiation into mature bone [3, 10]. The softened bone associated with FD likely led to encephalocele formation in our patients. There are a few case reports of epilepsy in patients with FD of the skull [4-9]. However, encephaloceles were not identified, and in one case there was speculation that seizures were secondary to alteration in the mechanism of cAMP as a messenger of the cerebral cortex [4]. It is possible that encephaloceles may have been present and went unrecognized.

Temporal encephaloceles are an increasingly identified cause of drug-resistant epilepsy and may be missed on MRI without careful review [2]. T2 sequences may be helpful in identifying encephaloceles due to high contrast between cerebrospinal fluid and bone signals [2]; however, a skull base CT may provide higher resolution of the temporal bone contour. Drug-resistant epilepsy due to temporal lobe encephaloceles often responds to surgical repair, whether lesionectomy or ATLAH [1]. There is no evidence of superiority of these techniques, although examples such as Case 1 may push physicians to offer more extensive resection, depending on the dominant hemisphere and patient-specific factors.

Our cases suggest that FD of the skull may be associated with encephalocele formation and subsequent epilepsy. For patients with known FD and epilepsy, neurologic imaging should be closely reviewed to determine the presence of encephaloceles. Recognition is crucial for appropriate



■ **Figure 2.** Fibrous dysplasia with an encephalocele in Case 2. (A) Head CT demonstrates ground-glass expansion of the right temporal bone diploic space (arrow), classic for fibrous dysplasia. (B-D) MRI (axial, sagittal, and coronal sequences) shows herniation of brain tissue into the sphenoid wing defect, confirming the encephalocele (arrow).

management, as surgical treatment may be curative in appropriately selected cases. ■

Supplementary material.

Summary slides and supplementary figure accompanying the manuscript are available at www.epilepticdisorders.com.

Disclosures.

The authors have no relevant financial disclosures or conflicts of interest to disclose.

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TEST YOURSELF

(1) What is fibrous dysplasia?

(2) What is the relationship between encephaloceles and epilepsy?

(3) What is an appropriate treatment for drug-resistant epilepsy related to encephaloceles?

Note: Reading the manuscript provides an answer to all questions. Correct answers may be accessed on the website, www.epilepticdisorders.com.