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Orbital Encephalocele Due to Sphenoid Wing Dysplasia in Neurofibromatosis Type 1: A Case Report Highlighting a Diagnostic Pitfall

Case Report

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Abstract

Neurofibromatosis type 1 (NF-1) is a multisystem genetic disorder characterized by cutaneous, neurological, and skeletal manifestations. Sphenoid wing dysplasia is a characteristic but relatively uncommon osseous abnormality in NF-1 and may result in herniation of intracranial contents into the orbit, producing an orbital encephalocele and progressive proptosis. In routine clinical practice, imaging evaluation of proptosis in NF-1 often prioritizes exclusion of optic pathway glioma or optic nerve sheath meningioma, which may lead to under-recognition of sphenoid wing dysplasia and its complications. Consequently, orbital encephalocele may be misdiagnosed as an arachnoid cyst.

This report describes two patients with NF-1—one pediatric and one adult—both presenting with unilateral proptosis and previously misdiagnosed as having arachnoid cysts. Through these cases, characteristic CT and MRI features facilitating accurate diagnosis are highlighted, emphasizing the importance of careful skull base assessment to avoid this important diagnostic pitfall.

Keywords: Neurofibromatosis type 1; Sphenoid wing dysplasia; Orbital encephalocele; Proptosis; Arachnoid cyst; Diagnostic pitfall

Neurofibromatosis type 1 (NF-1) is a common autosomal dominant neurocutaneous disorder caused by pathogenic variants in the *NF1* gene on chromosome 17, with an estimated incidence of approximately 1 in 3,000 individuals. The disease is characterized by marked phenotypic heterogeneity and involves multiple organ systems, including the skin, nervous system, and skeleton. While cutaneous manifestations and optic pathway gliomas are well recognized and frequently emphasized in clinical practice, skeletal abnormalities—despite being part of the diagnostic criteria—are comparatively under-recognized.

Sphenoid wing dysplasia is a characteristic but relatively uncommon skeletal manifestation of NF-1, reported in approximately 5–10% of affected individuals. Partial or complete absence of the sphenoid wing results in loss of the normal osseous barrier between the orbit and the middle cranial fossa, predisposing to herniation of cerebrospinal fluid (CSF) and brain parenchyma into the orbit. This may lead to the development of an orbital encephalocele, presenting clinically with progressive, often unilateral proptosis and potential optic nerve compression.

In routine imaging evaluation of proptosis in patients with NF-

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1, diagnostic attention is frequently directed toward excluding optic pathway glioma or optic nerve sheath meningioma, given their strong association with the disease. This diagnostic bias may result in under-recognition of sphenoid wing dysplasia and its complications. Furthermore, because orbital encephaloceles often demonstrate CSF-like signal characteristics on MRI, they may be misdiagnosed as arachnoid cysts, particularly when osseous abnormalities are subtle or not adequately assessed.

This case series describes two patients with NF-1—one adult and one pediatric—both presenting with unilateral proptosis and initially misdiagnosed as having arachnoid cysts. Through detailed CT–MRI correlation, this report highlights characteristic imaging features that facilitate accurate diagnosis, underscores an important diagnostic pitfall, and emphasizes the clinical and management implications of early recognition.

Ethical Considerations

Written informed consent was obtained from both patients and/or their legal guardians for publication of clinical details and imaging findings. All procedures were conducted in accordance with institutional ethical standards and the principles of the Declaration of Helsinki.

Case Series

Case 1

A 39-year-old man with a known diagnosis of neurofibromatosis type 1 presented with progressively increasing left-sided proptosis over several years. The patient reported mild visual discomfort but no acute visual loss, diplopia, or pain. There was no history of trauma or prior orbital surgery. Clinical examination revealed non-pulsatile left proptosis without signs of acute inflammation.

MRI of the brain and orbits with contrast demonstrated hypoplasia of the left sphenoid wing with herniation of brain parenchyma and CSF into the superior aspect of the left orbit, consistent with a left

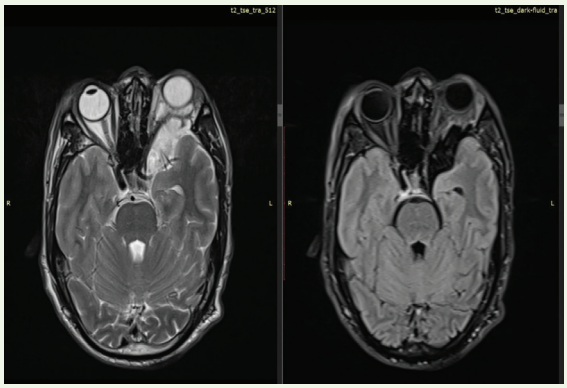

Figure 1: MRI brain, axial T2-weighted image OF 39-YEAR-OLD GENTLEMAN WITH KNOWN NF-1 demonstrates prominent arachnoid spaces in the left anterior temporal region, secondary to the orbital encephalocele. There is compression and medial deviation of the left optic nerve without abnormal enhancement, excluding optic nerve glioma or optic nerve sheath meningioma. There is severe narrowing of left superior orbital fissure and compression of optic nerve at this level.

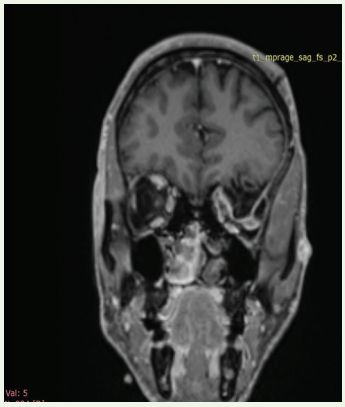

Figure 2: MRI brain, post contrast coronal T1 FS-weighted image OF 39-YEAR-OLD GENTLEMAN WITH KNOWN NF-1 demonstrates herniation of brain parenchyma and cerebrospinal fluid into the superior aspect of the left orbit, consistent with a left orbital encephalocele, resulting in left-sided proptosis.

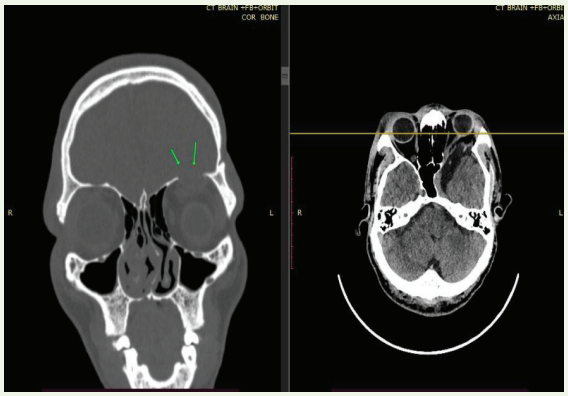

Figure 3: CT BRAIN (CORONAL AND AXIAL) OF 39-YEAR-OLD GENTLEMAN WITH KNOWN NF-1 demonstrates hypoplasia of the left sphenoid wing (arrow) with loss of the normal bony separation between the middle cranial fossa and the orbit, allowing herniation of intracranial contents into the left orbit.

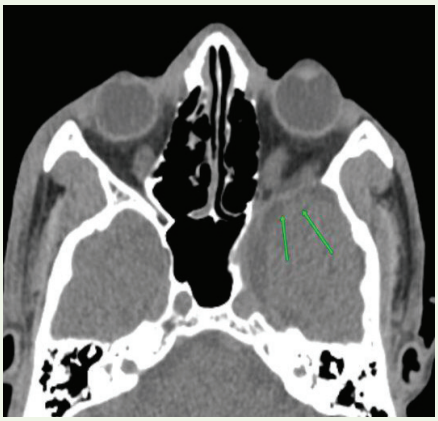

Figure 4: CT brain (axial bone window) of a 39-year-old man suspected to have NF-1 demonstrates dysplasia of the left sphenoid wing (arrow) with anterior extension of intracranial contents toward the posterior aspect of the left orbit.

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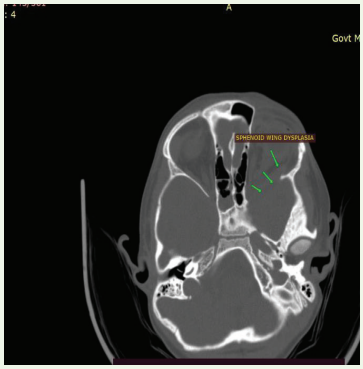

Figure 5: CT brain (axial bone window) of an 8-year-old boy suspected to have NF-1 demonstrates dysplasia of the left sphenoid wing (arrow) with anterior extension of intracranial contents toward the posterior aspect of the left orbit.

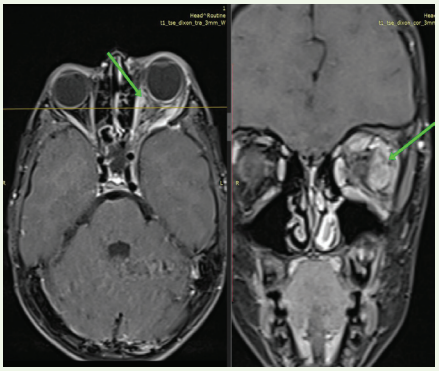

Figure 6: MRI orbit, axial and coronal post-contrast fat-suppressed T1-weighted image shows enhancing enlargement of the left medial and lateral rectus muscles (arrows) with mild surrounding orbital fat stranding, suggestive of extraocular muscle neurofibroma in the background of neurofibromatosis type 1.

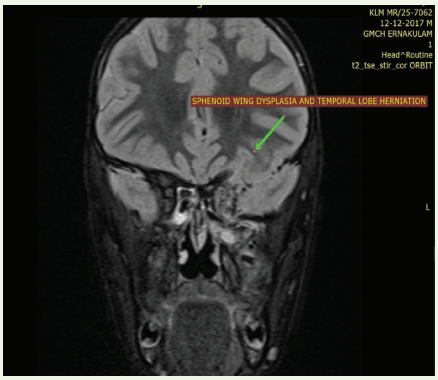

Figure 7: MRI orbit, coronal T2-weighted image of an 8-year-old boy suspected to have NF-1 demonstrates anterior herniation of the left temporal lobe into the posterior portion of the left orbit, consistent with an orbital encephalocele, resulting in left-sided proptosis and mild asymmetric enlargement of the left globe.

orbital encephalocele. This resulted in left-sided proptosis with compression and mild medial deviation of the intraorbital segment of the left optic nerve. Marked narrowing of the left superior orbital fissure was noted, with optic nerve compression at this level.

Prominent arachnoid spaces were seen in the left anterior temporal region secondary to the encephalocele, with medial displacement of the left middle cerebral artery. No abnormal enlargement or post-contrast enhancement of the optic nerve was identified, effectively excluding optic nerve glioma and optic nerve sheath meningioma. Additional findings included multiple scalp neurofibromas and T2/FLAIR hyperintense foci involving the bilateral thalami and periventricular regions, consistent with focal areas of signal intensity (FASI). The left sphenoid sinus was hypoplastic.

CT of the skull base confirmed hypoplasia of the left sphenoid wing with loss of the normal bony separation between the middle cranial fossa and the orbit. Prior imaging studies had described the lesion as an arachnoid cyst, leading to delayed recognition of the underlying pathology.

The patient was referred for multidisciplinary evaluation, and given the slowly progressive nature of symptoms and preserved vision, conservative management with clinical and imaging follow-up was advised.

Case 2

An 8-year-old boy, born to a mother with neurofibromatosis type 1, presented with gradually progressive left-sided proptosis noted by caregivers over the preceding year. There was no history of trauma, pain, or visual complaints. Ophthalmologic evaluation revealed mild asymmetry of the globes without acute visual deficit.

MRI of the brain and orbits with contrast, supplemented by CT imaging, revealed dysplasia of the left sphenoid wing with anterior herniation of the left temporal lobe into the posterior aspect of the left orbit, consistent with an orbital encephalocele. This resulted in left-sided proptosis and mild asymmetric enlargement of the left globe.

The left medial and lateral rectus muscles were enlarged and demonstrated post-contrast enhancement, suggestive of extraocular muscle neurofibromas, with mild surrounding orbital fat stranding. The intraorbital segment of the left optic nerve appeared mildly tortuous but showed no enlargement or abnormal enhancement. CT demonstrated absence of tram-track calcification, excluding optic nerve sheath meningioma. The left superior orbital fissure was asymmetrically enlarged, and the left sphenoid sinus was hypoplastic.

MRI of the brain demonstrated multiple non-enhancing T2/FLAIR hyperintense lesions involving the bilateral globus pallidi, thalami, cerebellum, and cerebral peduncles, characteristic of FASI. No diffusion restriction or mass effect was observed. As in the first case, the lesion had previously been interpreted as an arachnoid cyst.

Given the patient's age and absence of visual compromise, a conservative approach with close ophthalmologic and radiologic surveillance was recommended.

Discussion

Sphenoid wing dysplasia is a distinctive skeletal manifestation of neurofibromatosis type 1 and represents an important cause of

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unilateral proptosis in this population. Although included in the diagnostic criteria for NF-1, it remains relatively uncommon and is therefore frequently overlooked in routine imaging practice. The underlying pathogenesis is not fully understood but is thought to involve abnormal mesodermal development, progressive osseous remodeling, or pressure erosion related to adjacent neurofibromatous tissue.

Loss or hypoplasia of the sphenoid wing results in widening of the superior orbital fissure and disruption of the normal barrier between the orbit and middle cranial fossa. This permits herniation of CSF and brain parenchyma into the orbit, forming an orbital encephalocele. Clinically, this manifests as progressive or pulsatile proptosis and may lead to optic nerve compression, visual impairment, and cosmetic deformity.

Both cases in this series illustrate a common and clinically significant diagnostic pitfall: misinterpretation of orbital encephalocele as an arachnoid cyst. While both entities demonstrate CSF-like signal intensity on MRI, several imaging features favor encephalocele, including continuity of herniated brain tissue with intracranial structures, associated sphenoid wing dysplasia on CT, displacement of adjacent vascular structures, enlargement or narrowing of the superior orbital fissure, and the presence of other NF-1–related findings such as FASI and neurofibromas.

This case series adds to existing literature by emphasizing that reliance on MRI alone may be insufficient and that CT–MRI correlation is essential for accurate diagnosis. Prior reports have similarly highlighted that failure to assess the skull base can delay diagnosis and appropriate management. Early recognition is clinically relevant, as progressive orbital encephaloceles may require surgical reconstruction to prevent irreversible visual loss and significant cosmetic deformity.

From a management perspective, asymptomatic or slowly progressive cases may be managed conservatively with close follow-up, while patients with visual compromise or rapidly progressive deformity may benefit from surgical intervention. Accurate diagnosis therefore directly influences clinical decision-making and patient outcomes.

Conclusion

Orbital encephalocele secondary to sphenoid wing dysplasia is an important but under-recognized manifestation of neurofibromatosis type 1 and a frequent diagnostic mimic of arachnoid cyst. In NF-1 patients presenting with unilateral proptosis, exclusive focus on optic nerve tumors may lead to misdiagnosis. Careful evaluation of skull base anatomy with CT–MRI correlation is essential for accurate diagnosis, appropriate management, and prevention of visual morbidity.

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