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Sudden visual loss revealing a SMARCB1-deficient sinonasal undifferentiated carcinoma: A case report

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Abstract

Background: Sinonasal undifferentiated carcinoma (SNUC) is a rare and aggressive malignancy of the paranasal sinuses. Among its molecular subtypes, SMARCB1-deficient SNUC represents a particularly aggressive variant with poor prognosis and distinct diagnostic and therapeutic challenges.

Case presentation: We report a case of a 28-year-old male with no prior medical history who [4] presented with sudden right-sided visual acuity loss, associated with periorbital pain and frontal headaches. Ophthalmologic examination revealed a stage 3 papilledema. Urgent MRI showed a large ethmoid-centered mass with intracranial extension and mass effect. An endonasal biopsy confirmed the diagnosis of SMARCB1-deficient SNUC, staged T4aN0M0. The patient was managed with neoadjuvant cisplatin-etoposide chemotherapy followed by concurrent chemoradiotherapy. A post-treatment evaluation with FDG PET-CT is planned.

Conclusion: This case highlights the importance of considering sinonasal malignancies in the differential diagnosis of acute visual symptoms. SMARCB1-deficient SNUC requires early diagnosis, molecular confirmation, and aggressive multimodal management, ideally in expert centers following established guidelines.

Keywords: Sinonasal undifferentiated carcinoma; SMARCB1-deficient; Visual loss; Orbital symptoms; Paranasal sinus tumor; Case report.

Introduction

Sinonasal undifferentiated carcinoma (SNUC) is a rare epithelial malignancy, accounting for less than 5% of all sinonasal tumors. It is characterized by aggressive behavior, rapid local progression, and poor prognosis [1,2]. Among the recently identified molecular subtypes, SMARCB1-deficient SNUC has emerged as a distinct entity. SMARCB1 (INI1) is a tumor suppressor gene encoding a component of the SWI/SNF chromatin remodeling complex [3]. Loss of SMARCB1 expression is associated with dedifferentiation and resistance to conventional treatment [4]. Patients often present at an advanced stage with non-specific symptoms such as nasal obstruction, epistaxis, or cranial nerve deficits. Visual impairment as the initial presenting feature is uncommon and reflects rapid tumor invasion of the orbit or anterior skull base [5]. We present a rare case of SMARCB1-deficient SNUC revealed by sudden unilateral visual loss, illustrating the diagnostic and therapeutic challenges posed by this entity.

Case presentation

A 28-year-old male with no prior medical history presented to the ophthalmology emergency department with sudden visual acuity loss in the right eye, associated with right periorbital pain and right-sided frontal headaches. He reported no nausea or vomiting. Ophthalmologic examination revealed a best-corrected visual acuity of 0.5 in the right eye (OD) and 10/10 in the left eye (OS). Slit-lamp examination was unremarkable in both eyes. Fundoscopy revealed a stage 3 papilledema in the right eye. Given the acute onset and concerning fundoscopic findings, an urgent cerebral and orbital MRI was performed (Figure 1). The MRI revealed a large ethmoid-centered tumor measuring approximately 4.5 cm in the transverse plane and 6 cm in the coronal plane. The mass exhibited symmetrical intracranial extension into the anterior cranial fossa, elevating the orbital surfaces of the frontal lobes and causing extensive vasogenic edema. A mass effect was observed on the corpus callosum and anterior horns of the lateral ventricles. The tumor displaced the lateral walls of the ethmoid sinuses, without direct intraorbital or intracanalicular invasion, although narrowing of the optic canals was noted. The lesion extended into both nasal cavities, reaching below the level of the inferior turbinate on the left side. Additional findings included mucosal retention within the frontal and sphenoidal sinuses. The sella turcica and cavernous sinuses were preserved (Figure 2). An endonasal biopsy confirmed the diagnosis of SMARCB1-deficient sinonasal undifferentiated carcinoma, classified as T4aN0M0 according to the AJCC staging system. A multidisciplinary tumor board was convened. The patient was scheduled to undergo neoadjuvant chemotherapy with cisplatin and etoposide, followed by concurrent chemoradiotherapy based on cisplatin. A post-treatment evaluation with FDG PET-CT was planned to assess therapeutic response.

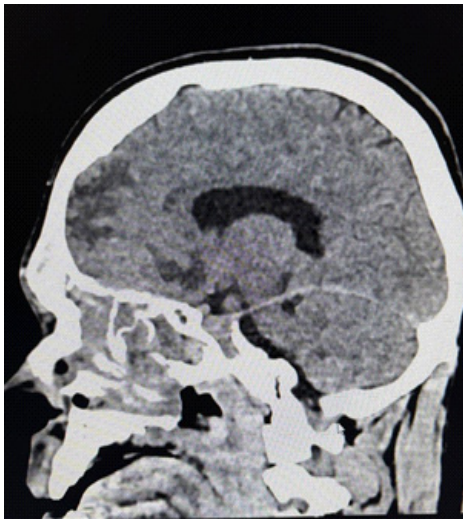

Figure 1: Sagittal CT scan of the brain and paranasal sinuses showing a large ethmoid-centered mass with anterior cranial fossa extension.

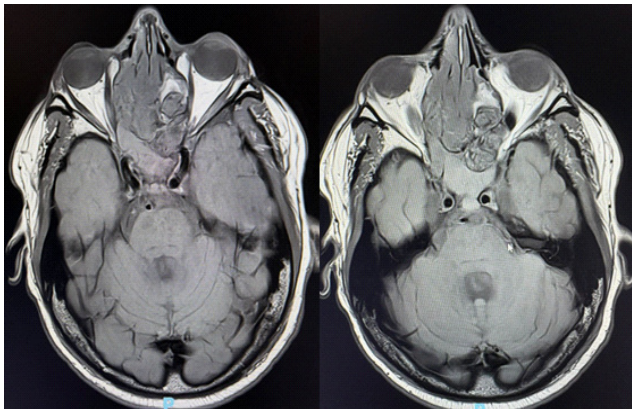

Figure 2: Axial T1-weighted MRI images with gadolinium contrast, showing a large ethmoid-centered tumor with heterogeneous enhancement. The mass invades the nasal cavity, right orbit, and erodes the cribriform plate, with anterior cranial fossa extension.

Discussion

Sinonasal undifferentiated carcinoma (SNUC) is a rare and highly aggressive malignancy, first described in the 1980s, arising from the epithelial cells of the nasal cavity and paranasal sinuses. Among its molecular subtypes, the SMARCB1-deficient variant has recently emerged as a distinct clinicopathological entity [6], with specific diagnostic, prognostic, and therapeutic implications. SMARCB1 (INI1) is a tumor suppressor gene encoding a core subunit of the SWI/SNF chromatin remodeling complex. Its loss is associated with dedifferentiation, high-grade histology, and resistance to therapy. SMARCB1-deficient tumors have been well-described in rhabdoid tumors, epithelioid sarcomas, and atypical teratoid/rhabdoid tumors of the CNS. In the head and neck region, SMARCB1-deficient SNUC represents a highly aggressive form with dismal prognosis and a median overall survival often below 18 months [8].

Clinically, SMARCB1-deficient SNUC often presents at an advanced stage with local invasion of the orbit, skull base, or intracranial compartment. Visual symptoms, while uncommon as an initial presentation, may reflect early orbital compression or intracranial spread, as was the case in our patient.

MRI findings typically reveal an ethmoid-centered mass with aggressive local behavior, vasogenic edema, and mass effect, highlighting the rapid progression of the disease [9].

Diagnosis requires histopathologic confirmation with immunohistochemistry. SMARCB1-deficient tumors characteristically show a complete loss of nuclear expression of INI1 protein. This feature, in combination with undifferentiated histology and sinonasal location, is sufficient for diagnosis. Differential diagnoses include other high-grade sinonasal tumors such as olfactory neuroblastoma (high-grade), neuroendocrine carcinoma, NUT carcinoma, and lymphoma.

According to REFCOR guidelines, the management of SNUC should be multidisciplinary and centralized in experienced centers. Neoadjuvant chemotherapy followed by surgery and/or radiotherapy remains the current standard, especially for locally advanced tumors. The recommended neoadjuvant regimen typically includes platinum-based chemotherapy, aiming to reduce tumor volume and assess chemosensitivity. In selected cases, complete surgical resection followed by adjuvant radio-

therapy may improve local control.

The prognosis of SMARCB1-deficient SNUC remains poor despite aggressive multimodal therapy. The identification of this molecular subtype raises the possibility of targeted therapies, including epigenetic modulators or immune checkpoint inhibitors, although clinical evidence is still limited [10]. This case highlights the importance of early recognition of unusual symptoms such as visual loss, prompt imaging, and thorough histopathological analysis, including molecular profiling. Given the aggressive nature of SMARCB1-deficient SNUC, early referral to a specialized center and adherence to national guidelines like REFCOR are essential for optimizing patient outcomes.

Conclusion

SMARCB1-deficient sinonasal undifferentiated carcinoma is a rare but highly aggressive malignancy with a poor prognosis and distinct molecular characteristics. Its initial presentation can be misleading, as illustrated by our case where sudden visual loss was the first and only symptom leading to diagnosis. This highlights the importance of considering sinonasal tumors in the differential diagnosis of acute neuro-ophthalmologic symptoms.

Early imaging, prompt histological and immunohistochemical evaluation are crucial for timely diagnosis. Given the aggressive course of SMARCB1-deficient SNUC, early multidisciplinary management, ideally in referral centers following national guidelines such as those provided by REFCOR, is essential. Further studies are needed to better define targeted therapeutic options and improve outcomes in this challenging and rapidly progressive disease.

Declaration of conflicts of interest: The authors declare no potential conflicts of interest.

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