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Palpebral Swelling Revealing Right Spheno-Orbital Aplasia in the Context of Neurofibromatosis Type 1: Report of A Rare Case

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Abstract **Case Report**

Neurofibromatosis type 1 (NF1) is an autosomal dominant genetic disorder frequently associated with skeletal abnormalities and cranio-orbital dysplasias. Orbital involvement is rare but may lead to cosmetic, neurological, and functional complications. We report an unusual case of a right palpebral hemangioma overlying aplasia of the greater wing of the sphenoid, in the context of NF1. A 17-year-old male with a history of NF1 was referred to radiology for evaluation of a stable right palpebral mass present since childhood. Clinical examination revealed a painless swelling, with no visual deficit or compressive signs. Facial CT scan demonstrated a well-defined mass in the right lateral can thus, measuring 46 × 16 mm, with homogeneous enhancement, consistent with a hemangioma. Imaging also revealed complete aplasia of the right greater wing of the sphenoid, meningeal herniation into the orbit, displacement of the lateral rectus muscle and optic nerve, and orbito-zygomatic bone dysmorphia. This constellation falls within the spectrum of NF1 orbital anomalies, in which sphenoidal aplasia, though rare, is highly suggestive. The coexistence of a benign vascular tumor such as a hemangioma, although nonspecific, raises the question of abnormal vascularization in NF1-affected tissues. This case highlights an atypical orbital presentation of NF1. Imaging plays a crucial role in establishing the diagnosis, guiding management, and anticipating potential complications.

Keywords: Neurofibromatosis type 1, Sphenoidal aplasia, Palpebral hemangioma, Facial CT.

INTRODUCTION

Neurofibromatosis type 1 (NF1), or Von Recklinghausen's disease, is a multisystemic genetic disorder with heterogeneous clinical manifestations. In addition to the characteristic cutaneous signs (café-au-lait macules, Lisch nodules, neurofibromas), certain osseous lesions, though less frequent, are highly specific and occasionally revealing.

Among them, aplasia of the greater wing of the sphenoid is a rare cranio-orbital anomaly, which may result in pathological widening of the superior orbital fissure, meningeal protrusion, and orbital bone remodeling. The association with a palpebral vascular mass, though rarely described, may fit within the framework of NF1-related vascular abnormalities.

CASE PRESENTATION

A 17-year-old adolescent, with a confirmed diagnosis of NF1, was referred for radiological evaluation of a right palpebral swelling stable since childhood. No visual symptoms were reported. Clinical examination revealed a soft, non-pulsatile mass at the lateral canthus, without proptosis or oculomotor restriction.

Imaging (Contrast-enhanced CT):

- Right soft tissue mass, 46 × 16 mm, well-circumscribed, showing homogeneous post-contrast enhancement: consistent with a hemangioma (Figure 1).
- Complete absence of the right greater wing of the sphenoid, with widening of the superior orbital fissure (Figure 2).
- Herniation of the dura mater into the orbital space, mildly displacing the lateral rectus muscle and optic nerve (Figure 3).
- Dysplasia of the frontal zygomatic process (Figure 4).
- Bilateral mucosal thickening of the maxillary sinuses, without osteolysis.

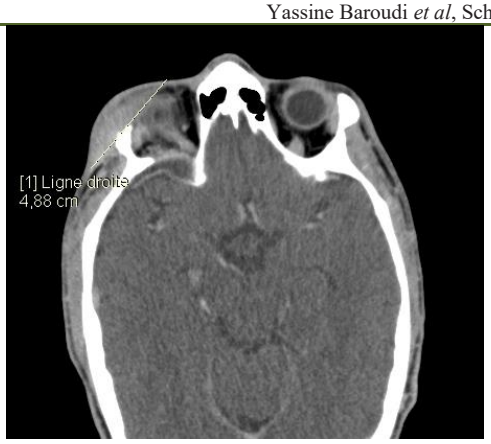

Figure 1: Axial CT scan (soft tissue window, post-contrast) showing a well-circumscribed soft tissue mass in the right lateral canthus with homogeneous enhancement, consistent with a hemangioma

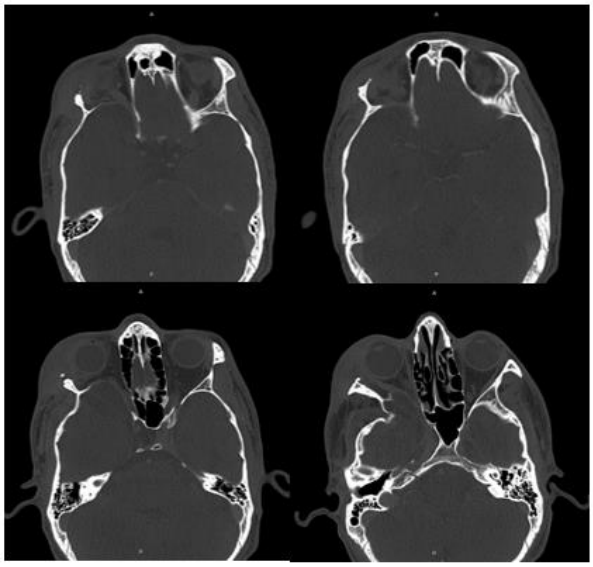

Figure 2: Axial CT scan (bone window) demonstrating complete aplasia of the right sphenoid greater wing with widening of the superior orbital fissure, compared to the normal left side

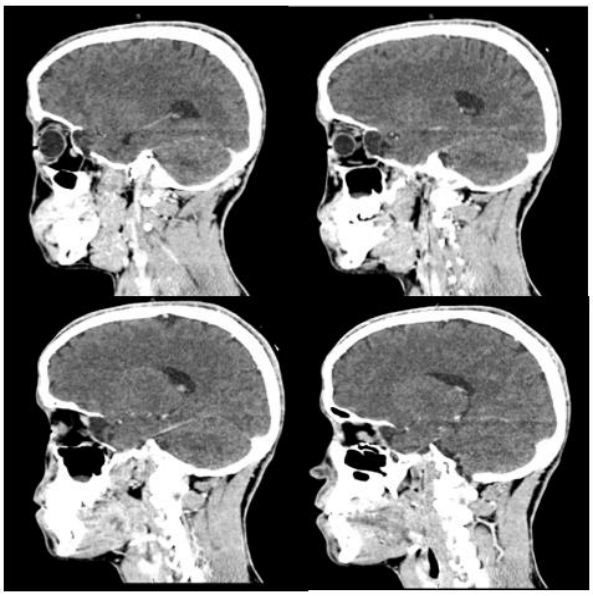

Figure 3: Sagittal CT scan (soft tissue window) showing herniation of the dura mater into the orbit through the sphenoid aplasia, displacing retrobulbar muscular structures

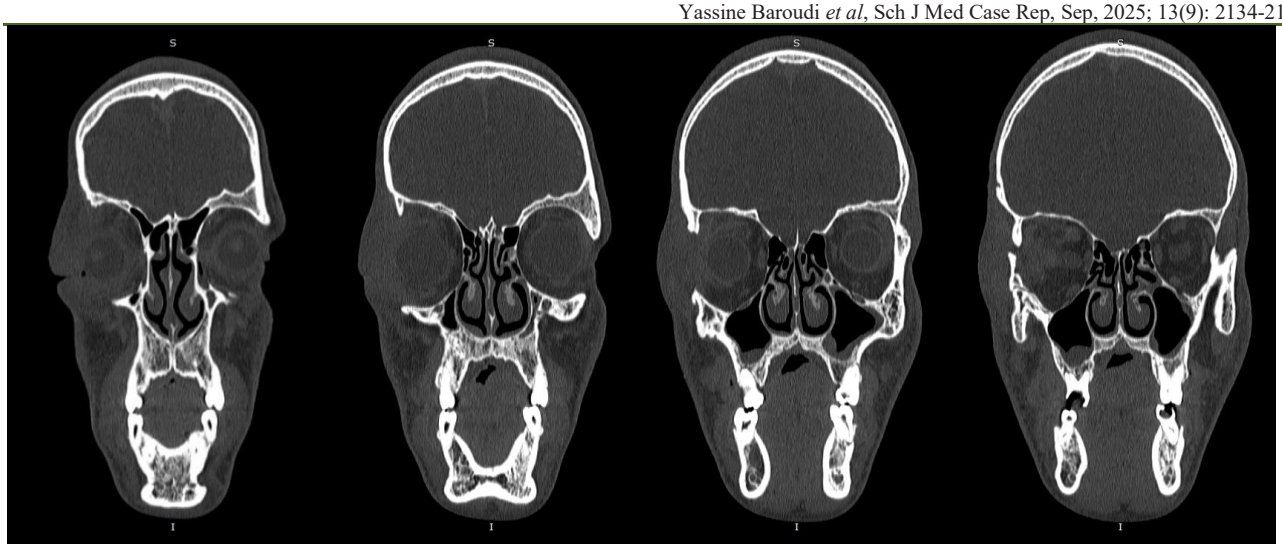

Figure 4. Coronal CT scan (bone window) showing dysmorphia of the right orbital framework with hypoplasia of the frontal zygomatic process, consistent with NF1-related orbitocranial anomaly

MANAGEMENT

Given the absence of functional signs, visual impairment, or proptosis, a conservative approach was adopted. Regular ophthalmological and radiological follow-up was recommended to monitor the palpebral mass and assess for possible functional repercussions.

An angiographic evaluation and embolization were considered but deferred, owing to the lesion's stable, painless, and non-progressive nature.

DISCUSSION

Orbital bone abnormalities in NF1 are rare but well documented, most often in the form of aplasia or hypoplasia of the greater wing of the sphenoid. This anomaly, although inconstant (5–10% of cases), is a minor diagnostic criterion in the NIH classification of NF1 [1].

Sphenoidal aplasia results in pathological widening of the superior orbital fissure, which may lead to herniation of intracranial contents (meningocele or encephalocele) into the orbit. Clinical manifestations may include proptosis, visual disturbance, or—as in our case—slowly progressive palpebral swelling. This finding, though nonspecific, is highly suggestive in a patient with NF1.

The association of a palpebral hemangioma with this osseous anomaly is rare. While benign vascular tumors are common in pediatric populations, their co-occurrence with orbital malformations in NF1 remains poorly documented. Some authors suggest that altered vascular microenvironments in NF1-affected tissues may promote endothelial proliferation [2].

Imaging, particularly contrast-enhanced CT and MRI, plays a pivotal role in evaluating these complex lesions by: Precisely identifying the origin of the tumoral process,

- Defining its extension to adjacent structures (muscles, nerves, meninges),
- And guiding therapeutic strategies.

In our case, CT imaging revealed not only the palpebral hemangioma but also a major cranio-orbital malformation with displacement of the lateral rectus muscle and optic nerve, though without acute compression.

Management depends on functional and cosmetic impact. A conservative approach may be justified in the absence of visual impairment or rapid growth. Conversely, preoperative embolization should be considered in symptomatic large hemangiomas or surgical candidates, as it reduces intraoperative bleeding risks, particularly in the context of abnormal vascularization [3].

Finally, this case emphasizes the importance of multidisciplinary follow-up in NF1. Collaboration among radiologists, ophthalmologists, neurosurgeons, and geneticists is essential to:

- Monitor the evolution of orbital lesions,
- Anticipate potential complications (optic compression, orbital deformity),
- And guide personalized therapeutic decisions.

CONCLUSION

This case underscores the diagnostic value of meticulous radiological assessment in atypical forms of NF1. The coexistence of sphenoidal aplasia with a palpebral vascular mass requires multidisciplinary evaluation involving radiology, neurosurgery, and ophthalmology. Imaging not only characterizes the lesion but also guides therapeutic management.

Conflicts of Interest: None declared.

Consent

- Informed consent was obtained from the patient and/or legal guardian for publication of this case and accompanying images.

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