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Orbital Neurofibroma Complicated by Grade III Exophthalmos in a Patient with Neurofibromatosis Type 1: A Case Report

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Abstract | **Case Report**

Introduction: Neurofibromatosis type 1 (NF1) is a genetic disorder predisposing individuals to nerve sheath tumors. Orbital involvement presenting as a large optic nerve neurofibroma in adults is a rare clinical entity that requires precise radiological characterization. ***Case Report:*** We report the case of a 60-year-old patient, followed for NF1, presenting with progressive right-sided proptosis. Orbital magnetic resonance imaging (MRI) revealed a right intra- and extraconal oculo-orbital mass causing grade III exophthalmos. The lesion demonstrated a heterogeneous intermediate signal on T1, T2, and SP FLAIR sequences, with heterogeneous contrast enhancement, confirming the diagnosis. ***Conclusion:*** Multi-sequence MRI is essential for mapping the extension of these orbital tumors, assessing their mechanical impact, and ruling out malignant transformation.

Keywords: Neurofibromatosis type 1; Optic nerve; Neurofibroma; Orbit; Exophthalmos; MRI.

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INTRODUCTION

Neurofibromatosis type 1 (NF1), or von Recklinghausen's disease, is a polymorphic autosomal dominant phakomatosis with cutaneous, neurological, and ophthalmological manifestations. It is caused by mutations in the NF1 tumor suppressor gene located on chromosome 17q11.2, which encodes neurofibromin.

Ophthalmologically and orbitally, the most classic manifestations in children include optic pathway gliomas and iris hamartomas (Lisch nodules). Orbital neurofibromas, whether plexiform or localized, represent another characteristic manifestation that may occasionally present or progress late in adulthood. Although benign in the vast majority of cases, their volume and tight anatomical localization within the orbital cavity can induce major mechanical and functional complications, warranting high-resolution imaging exploration.

CLINICAL CASE

We report the case of a 60-year-old patient, known and followed for neurofibromatosis type 1, referred for the evaluation of a progressive right-sided unilateral exophthalmos.

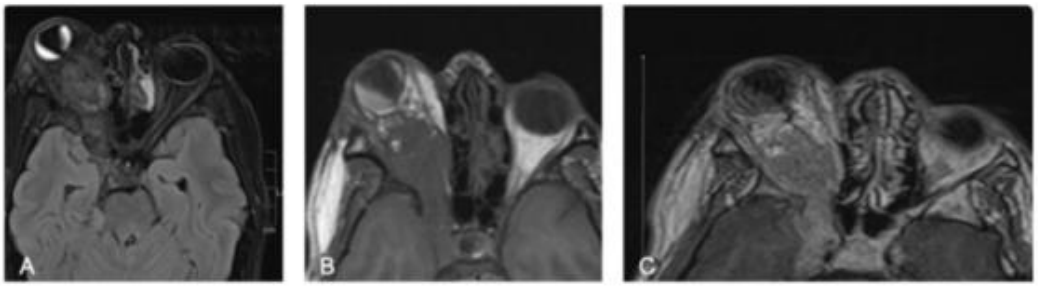
Magnetic Resonance Imaging (MRI) Evaluation

Orbital MRI was performed using a multi-sequence protocol including SP FLAIR (A), unenhanced T1-weighted (B), and contrast-enhanced T1-weighted (C) acquisitions. The semiological analysis revealed:

- **Topography and Extension:** A large oculo-orbital mass simultaneously occupying the intra- and extraconal compartments of the right orbit.
- **Mechanical Impact:** Anterior displacement of the globe resulting in severe ipsilateral grade III exophthalmos.
- **Signal Characteristics:** A mass with heterogeneous intermediate signal intensity across T1, T2, and SP FLAIR sequences, reflecting a complex tissue architecture.
- **Vascular Profile:** A pronounced yet heterogeneous enhancement of the mass following the administration of intravenous contrast.

The integration of clinical data (history of NF1) and MRI semeiology established the diagnosis of an optic nerve neurofibroma in the context of neurofibromatosis type 1.

Figure 1: A 60-year-old patient followed for neurofibromatosis type 1. Orbital MRI in SP FLAIR (A), T1 (B), and contrast-enhanced T1 (C). Right intra- and extraconal oculo-orbital mass lesion responsible for ipsilateral grade III exophthalmos, displaying heterogeneous intermediate signal on T1/T2 and FLAIR, with heterogeneous enhancement following contrast injection. Optic nerve neurofibroma in the setting of neurofibromatosis type 1.



IRM orbitaire en SP FLAIR (A), T1 (B) et T1 après injection (C)

DISCUSSION

Radio-Anatomical Correlations

An optic nerve neurofibroma or neurofibroma of the optic nerve sheath is a benign tumor originating from Schwann cells and perineurial fibroblasts. Unlike low-grade optic gliomas (pilocytic astrocytomas) predominantly seen in children, neurofibromas tend to spread along nerve pathways and infiltrate orbital fat spaces.

On MRI, the heterogeneous intermediate signal appearance across T1, T2, and FLAIR sequences reflects the mixed histological composition of the lesion, combining dense cellular bundles with areas rich in myxoid, cystic, or collagenous stroma. Heterogeneous contrast enhancement reflects the irregular vascularity of the tumor stroma. The combined involvement of intra- and extraconal spaces explains the significant mass effect and the severity of the grade III exophthalmos observed in our patient.

Differential Diagnosis

Given an evolving orbital mass in a 60-year-old adult with NF1, several differential diagnoses must be considered:

1. **Adult Optic Pathway Glioma:** A much rarer entity than in children, known for a significantly more aggressive biological and clinical course.
2. **Orbital Schwannoma:** Generally better defined, encapsulated, frequently showing central cystic degeneration and a target-like appearance on T2-weighted imaging.
3. **Malignant Transformation into Malignant Peripheral Nerve Sheath Tumor (MPNST):** A formidable complication occurring in 8% to 13% of patients with NF1 over their lifetime. It should be suspected in the presence of rapid tumor growth, the onset of intense orbital pain, and MRI findings demonstrating extensive areas of non-enhancing necrosis or aggressive bone lysis of the orbital walls.

Therapeutic Management

Management of deep orbital neurofibromas in adults is challenging due to their infiltrative nature and close proximity to the neurovascular structures of the orbital apex. In the absence of radio-clinical signs of malignant transformation, close ophthalmological and radiological surveillance via serial MRI is favored. Surgical debulking or orbital decompression is discussed only in cases of major functional threat (direct optic nerve compression causing decreased visual acuity) or severe exposure keratopathy secondary to grade III exophthalmos.

CONCLUSION

This clinical case illustrates a late and voluminous orbital manifestation of neurofibromatosis type 1 in a 60-year-old patient. Multi-sequence orbital MRI remains the essential diagnostic tool for characterizing the lesion, establishing a precise map of its intra- and extraconal extension, assessing ocular impact, and enabling early detection of potential malignant transformation.

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