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CASE REPORT

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Craniofacial plexiform neurofibroma revealed by exophthalmos: A case report

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ABSTRACT

Plexiform neurofibromas (PNFs) are benign tumors that arise from the nerve sheaths and are a hallmark of neurofibromatosis type 1. These tumors can occur in various anatomical locations, including the craniofacial region, and pose unique diagnostic and management challenges. They are often characterized by specific imaging features on Magnetic Resonance Imaging. In this case report, we present the imaging findings of a craniofacial PNF, highlighting its characteristic features, and compare them with the existing literature on craniofacial PNFs.

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Plexiform neurofibromas; target sign; nerve; MRI

Introduction

Plexiform neurofibromas (PNFs) are one of the most common and debilitating presentations of neurofibromatosis type 1 (NF1). They account for substantial morbidity, disfigurement, functional impairment, and can be life-threatening. They are benign tumors that originate from nerve sheath cells or subcutaneous peripheral nerves and can involve multiple fascicles [1].

Due to their large size and complex location, PNFs—particularly those affecting the orbit and sometimes extending to the temporal region and face—carry a high risk of neurological and functional damage during resection. Surgical interventions are commonly postponed for as long as possible. These tumors, although rare, are disfiguring and potentially devastating complications of NF1 [2].

This observation highlights the characteristic imaging features of PNFs, which can effectively lead to the correct diagnosis at an early stage of investigation.

Case Report

The patient was a 30-year-old man with no specific pathological antecedents, who presented with symptoms of intracranial hypertension, including headache, nausea, and blurred vision. These symptoms had progressively worsened over a 10-day period. Additionally, the patient developed progressive left exophthalmos, which further indicated a concerning neurological condition.

The patient's physical examination revealed multiple firm, non-tender, well-defined nodules in the frontotemporal region bilaterally. Notably, no café-au-lait spots were present. A slit-lamp examination of the right eye revealed Lisch nodules, characteristic of NF1 (Fig. 1). The neuro-ophthalmological examination showed normal visual acuity (20/20 in both eyes) and intact extraocular muscle movements. However, ophthalmoscopy revealed papilledema, with swelling of the optic disc indicating increased intracranial pressure.

On Magnetic Resonance Imaging(MRI), the T2 sequence revealed a heterogeneously hyperintense, infiltrating soft tissue tumor in the left fronto-parieto-occipital and infratemporal fossae, facial region, and right frontal soft tissue, characterized by a "bag of worms" appearance. The tumor is also located in the intra- and extra-conal fat of the left orbit, causing exophthalmos (Fig. 2). The lesions appear isointense on T1, with intense and heterogeneous

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Figure 1. The patient presents with left-sided exophthalmos, bilateral nodular lesions in the frontal region, and Lisch nodules in the right eye, consistent with findings seen in NF1.



Figure 2. MRI axial T2 sequence revealed a heterogeneously hyperintense, infiltrating, soft tissue tumor characterized by "bag of worms" appearance, located in the intra- and extra-conal fat of the left orbit, causing exophthalmos.

enhancement after gadolinium administration and no restriction on diffusion (Fig. 3).

Based on the diagnostic criteria for NF1 outlined in the 2021 international consensus, this patient meets several key features of NF1, including multiple neurofibromas (firm nodules in the frontotemporal region), **Lisch nodules** observed on slit-lamp examination, and an MRI showing a PNF with a "bag of worms" appearance. Although the patient lacks café-au-lait spots, the other findings, along with symptoms of intracranial hypertension and exophthalmos, strongly support the diagnosis of NF1.

Imaging features were consistent with a PNF. The patient has a positive genetic test for NF1. The histologic examination after mass biopsy revealed diffuse nerve fascicles distension caused by a proliferation of spindle-shaped cells embedded in myxoid matrix consistent with PNF.

Discussion

PNFs are complex nerve sheath tumors predominantly associated with NF1. These tumors can infiltrate multiple tissue planes, making their imaging and subsequent management particularly challenging. MRI is the reference standard modality for evaluating neural tissues. In PNFs, the tumor is typically lobulated, and the "target sign" on MRI is considered pathognomonic [1].

On T2-weighted MRI, the tumor usually shows high signal intensity, often described as a "bag of worms" appearance. The T2 hyperintensity reflects the mixture of different tissue types, such as fluid-filled cysts, myxoid stroma, and areas of necrosis. This heterogeneous appearance on T2-weighted images is one of the hallmarks of PNFs [3]. On T1-weighted MRI images, they typically appear as masses with low to intermediate signal intensity. After the administration of contrast agents, craniofacial PNFs often show heterogeneous enhancement, which is attributed to their complex structure [4].

Moreover, conventional MRI features, such as regular or irregular margins and T1W post-contrast heterogeneity, are helpful in differentiating benign from malignant peripheral nerve sheath tumors. Irregular margins and restricted diffusion

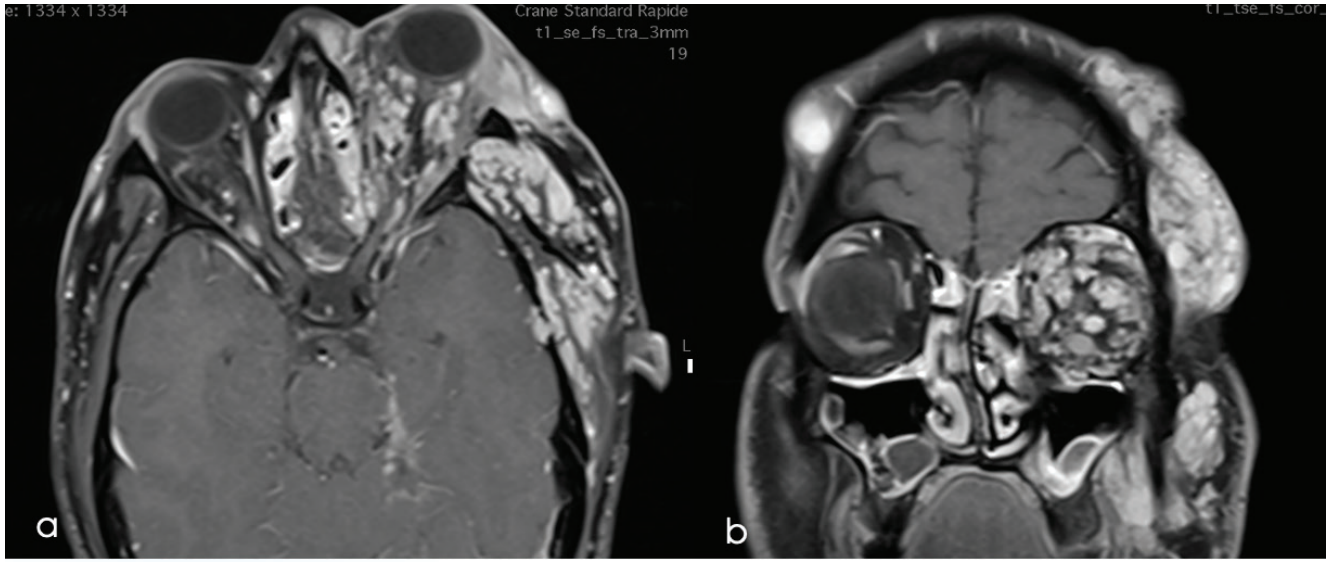


Figure 3. MRI axial and coronal T1 fat sat after injection of gadolinium objectified intense and heterogeneous enhancement of the lesions.

on diffusion-weighted imaging (DWI) sequences, along with the apparent diffusion coefficient (ADC) map, may suggest malignancy. These features, when incorporated into a conventional MRI examination, play a key role in distinguishing benign from malignant neurofibromas and can significantly affect clinical decision-making for patients with these tumors [5].

PNFs often exhibit a locally invasive growth pattern, extending into the surrounding tissues. This can involve adjacent muscles, skin, and bones. In some cases, the tumor may also extend into the orbit or sinuses, depending on its location within the craniofacial skeleton. It can cause a mass effect on adjacent structures. The tumors may lead to bone remodeling or erosion, especially when located in the paranasal sinuses, orbit, or skull base. In rare cases, PNFs may cause craniofacial deformities [6].

In cases of NF1 presenting with exophthalmos, it is crucial to consider optic pathway glioma (OPG) as a differential diagnosis. OPGs typically present as fusiform, well-defined tumors along the optic pathway with mild-to-moderate contrast enhancement, restricted diffusion on DWI, and lower ADC values, reflecting higher cellularity. They can involve the optic nerve, chiasm, and hypothalamus, and their growth is more localized. On the other hand, PNFs are less cellular, often showing higher ADC values, and grow expansively, affecting surrounding tissues [7].

The recommended treatment for PNFs is surgical resection, with a subtotal approach favored for debulking large tumors rather than total excision. This approach is preferred to minimize morbidity and prevent functional neurological impairment associated with complete tumor removal. In cases of unresectable or progressive tumors, the use of a Mitogen-Activated Protein Kinase Kinase 1 and 2 inhibitor has been explored as a potential treatment option [1].

Conclusion

PNFs are benign tumors that originate from nerve sheaths and are a hallmark of NF1. These tumors can occur in various anatomical locations, including the craniofacial region. MRI plays a crucial role in assessing the extent of tumor involvement and in planning surgical approaches, as resecting these tumors can be complicated by their proximity to critical structures.

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