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CASE REPORT

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A rare case of occipital plexiform neurofibroma with skull erosions in neurofibromatosis type 1

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Abstract

Plexiform neurofibromas are benign nerve sheath tumours commonly associated with neurofibromatosis type 1 (NF1). Plexiform neurofibromas of the scalp is a rare finding. Plexiform neurofibroma in the occipital region is even rarer. The commonest skull manifestations in neurofibromatosis involve the orbit, with very few reports about occipital defects. Magnetic resonance imaging (MRI) is the preferred modality to identify the lesion. It is usually managed by surgical excision, but recurrence has also been reported even after thorough removal. We present a case of a 52-year-old man with plexiform neurofibroma in the occipital region with skull erosions which is an extremely rare finding.

Keywords: plexiform neurofibroma, bone erosions, neurofibromatosis type 1 (NF1), occipital plexiform neurofibroma

Introduction

Neurofibromatosis type 1 (NF1) is an autosomal dominant disorder affecting 1 in 2500–3500 individuals. It has a penetrance of almost 100%. It is caused by mutations of the NF1 gene, which is located at chromosome 17q11.2. It is characterised by multiple neurofibromas, café-au-lait macules, and nervous system involvement. Plexiform neurofibromas, a hallmark feature of NF1, can lead to compressive neuropathies and neurological dysfunction. Plexiform neurofibromas are the least common variant and usually are pathognomonic for NF1. Plexiform neurofibromas of the scalp is a rare finding and its involvement in the occipital region is even rarer with a very few case reports about occipital defects. Magnetic resonance imaging (MRI) is the preferred modality to identify the lesion. One of the life-threatening complications is the malignant transformation of plexiform neurofibromas, and surgical intervention in NF-1-related osseous lesions is indicated for pain relief, cosmetic concerns, and neurological compromise.

We present a case of a 52-year-old man who developed acute right-sided limb weakness, ultimately diagnosed with plexiform neurofibroma with skull erosions.

Case presentation

A 52-year-old man presented with right upper limb and lower limb weakness for one day. There were no associated symptoms like headache, nausea, vomiting, chest pain, or palpitations. There was no

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prior diagnosis of NF1, and no history of similar neurological events. A large soft mass on the scalp in the occipital region had been present for over 15 years. He had a family history of NF. He was newly diagnosed with hypertension.

On general examination, there were multiple neurofibromas, café-au-lait macules, and a right-sided scalp swelling suggestive of a neurofibroma (6.0 cm x 3.0 cm x 11.0 cm) (Figure 1A and 1B). On the right upper and lower limbs with normal tone, reflexes and sensation. There was a right-sided upper motor neuron (UMN) facial nerve palsy. Vision and Fundoscopy was normal and Lisch nodules were not identified.

NCCT brain revealed small vessel disease with an infarction in the left corona radiata and a right-sided occipital soft tissue lesion with bony erosions (Figure 2A and 2B).

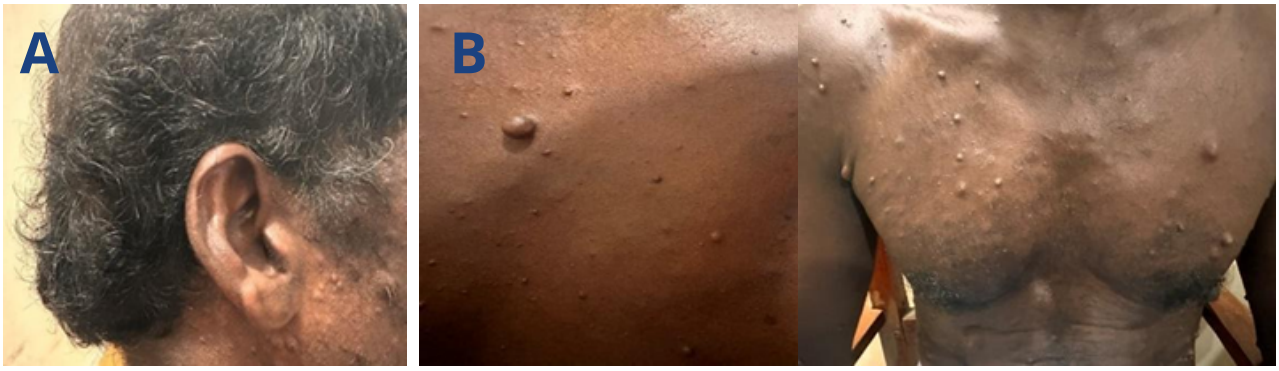

Figure 1. **(A)** Clinical photographs showing the right-sided occipital swelling. **(B)** Clinical photographs showing multiple neurofibromas on the chest

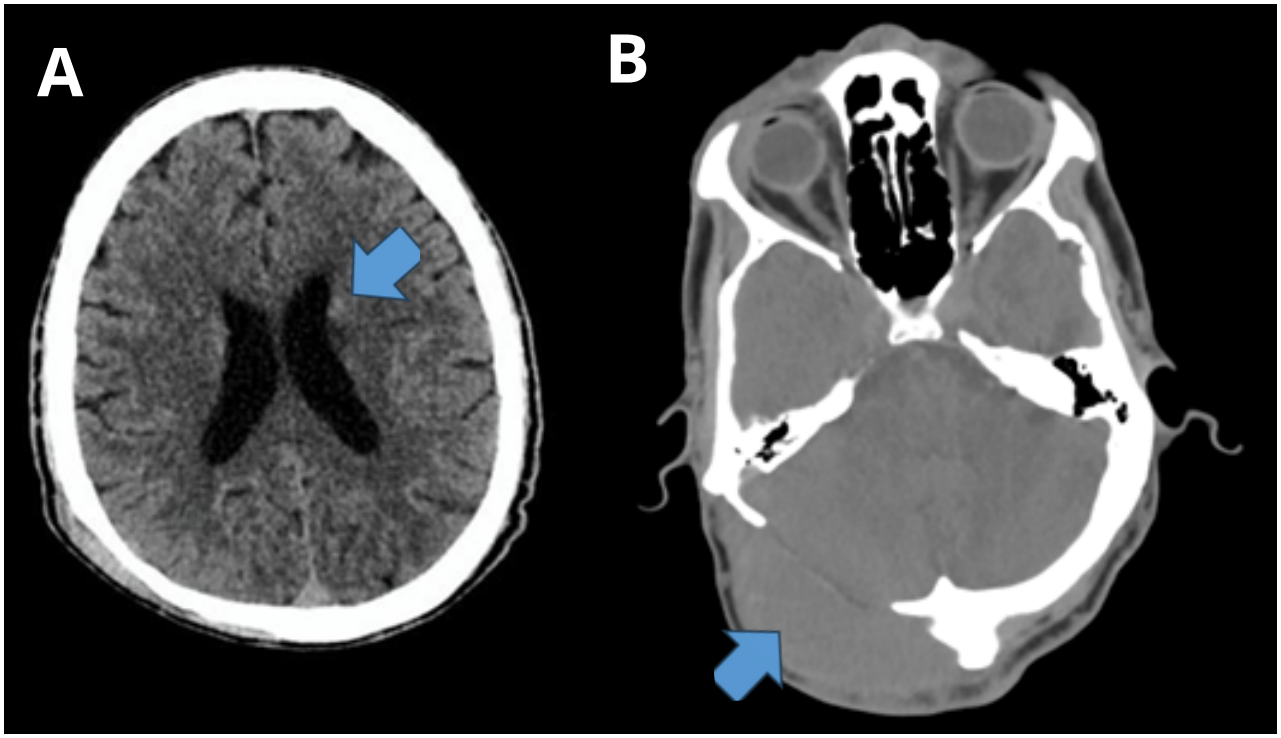

Figure 2. **(A)** NCCT brain revealed small vessel disease with an infarction in the left corona radiata. (marked by an arrow) **(B)** Right-sided occipital soft tissue lesion with bony erosions (marked by an arrow)

Magnetic Resonance Imaging (MRI) of the brain (Figure 3A), revealed a large lesion in the subcutaneous tissue plane in the right occipital region measuring 6.0 cm x 3.0 cm x 11.0 cm (anteroposterior x transverse X superior-inferior) associated with erosion of underlying bone. There was no infiltration of the sigmoid or transverse sinuses. The lesion is hypointense in T1-weighted images and heterogeneously high signal in T2-weighted images, with mild contrast enhancement. Only a few lesions showed a typical target sign in T2-weighted fat-suppressed images. Few foci of blooming artefacts were seen in susceptibility weighted imaging (SWI). There were no areas of diffusion restriction. There was no intra cerebral extension. Few other similar lesions were seen in the scalp in the left parietal region and in the frontal region. The overall picture was of a plexiform neurofibroma of the right occipital region with multiple scalp neurofibroma with underlying bone erosions. Old lacunar infarction with gliosis was seen in the left corona radiata.

Pheochromocytoma associated with NF was excluded by a urine VMA of 8.5 mg in 24 hours (1 to 11 mg).

Surgery was not performed on our patient as there was no neurological compromise or cosmetic concern due to the plexiform neurofibroma. Genetic counseling was offered to the patient and family members. Serial neuroimaging was arranged to monitor progression of the lesion.

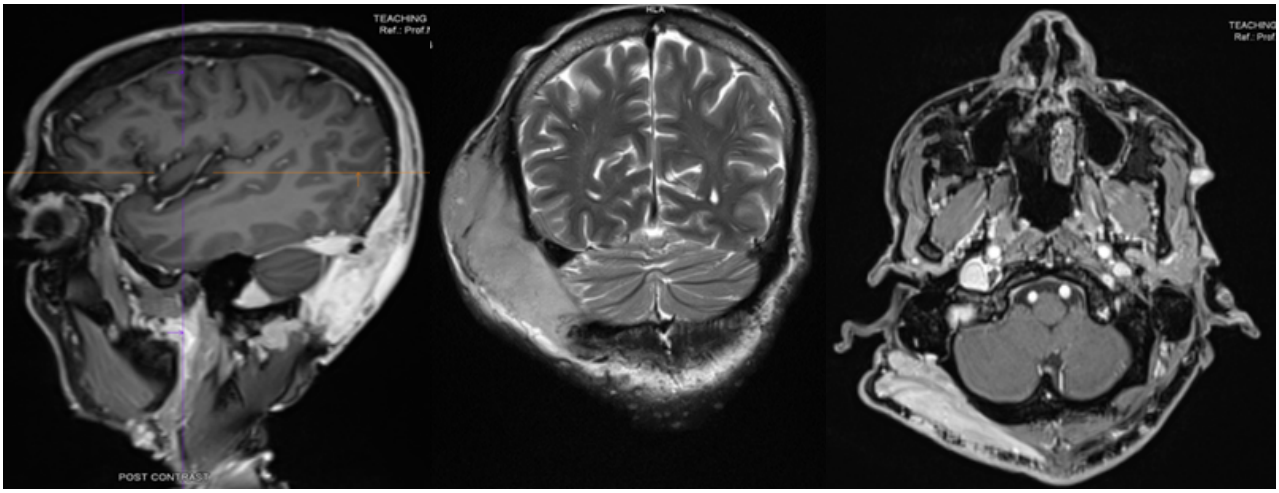

Figure 3. (A) MRI brain with contrast showing a large mass lesion seen in the subcutaneous tissue plane in the right occipital region

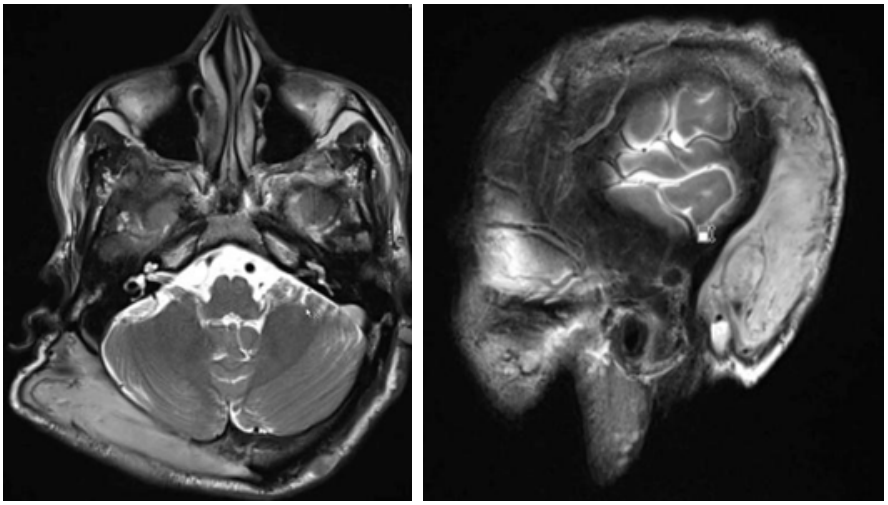

Figure 3. (B) MRI brain with contrast showing target sign in T2-weighted fat-suppressed images

Discussion

Plexiform neurofibromas are benign but can cause significant morbidity due to their infiltrative nature. Plexiform neurofibromas occur in only 17% of cases of NF 1, and the majority are solitary lesions (91%) and may undergo malignant transformation in 10-15% of cases. The commonest skull manifestations in neurofibromatosis involve the orbit. Plexiform neurofibroma involving the scalp is very rare and it involving the occipital region is extremely rare. Only a handful of cases have been reported in the medical literature worldwide which emphasises the limited occurrence.(1-4)

The criteria for diagnosis of NF1 has been introduced by the National Institutes of Health (NIH), according to which two or more criteria should be fulfilled for the diagnosis.(5) Our patient had two or more neurofibromas, one plexiform neurofibroma and a distinctive osseous lesion which fulfilled the diagnostic criteria for neurofibromatosis.

Neurofibromatosis Type 1 (NF-1) is primarily considered a neurocutaneous disorder of neural crest origin, with limited emphasis on osseous abnormalities. But skeletal dysplasia is one of the seven diagnostic criteria for NF-1, and osseous manifestations occur in up to half of the affected individuals. These skeletal changes are mainly thought to be due to the altered function of the NF-1 gene, with secondary bone involvement often resulting from tumour compression. Among these manifestations, cranial osteolysis is an extremely rare scenario, with only a few reported cases. The aetiology of cranial osteolysis is still unclear, with uncertainty whether it arises as a direct feature of NF-1 or as a secondary effect of tumour erosion.

In our patient, the calvarial erosion was likely secondary to chronic pressure from the overlying plexiform neurofibroma rather than primary skeletal dysplasia, as described in previous reports. Occipital bone involvement has been reported only rarely in NF1, making this case a clinically significant variant.
Ultrasound, computed tomography (CT), and magnetic resonance imaging (MRI) are useful in the diagnosis and characterisation of plexiform neurofibromas. However, MRI is the most useful imaging modality to characterise tumour size and extent.(6)

Surgery is offered in the case of plexiform neurofibromas when they cause disfigurement and malignant transformation. However, the recurrence rate is 20%despite an appropriate excision.(1)

In NF-1-related osseous lesions, surgical intervention is indicated for cosmetic concerns, pain relief, and neurological involvement. However, the surgical intervention of plexiform neurofibromas poses a considerable challenge due to the high vascularity of these tumours, which may end up in massive bleeding. Given these risks, some authors recommend limiting intervention to biopsy rather than extensive surgical excision. Further studies are necessary to improve the understanding of osseous involvement in NF-1 and to develop safer, more effective surgical strategies.

Surgical intervention should be individualized, particularly in large, highly vascular, or infiltrative tumors where operative risk and recurrence remain significant. There was no indication for surgery in our patient.

Conclusion

Occipital plexiform neurofibroma with associated calvarial erosion is an exceptionally rare manifestation of Neurofibromatosis type 1. This case highlights the importance of recognising plexiform neurofibromas and associated distinctive osseous lesions, as they are key components of the diagnostic criteria for NF1 and may present with unusual anatomical involvement.

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Declarations

Conflicts of interest:
The authors declare that they have no conflicts of interest with respect to the research, authorship, and/or publication of this article.

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