Atypical cavernous hemangioma clinically masquerading as a plexiform Orbitofacial neurofibromatosis: A case report

Sandip K Sahu, Sunil K Rout[1], Ranjit K Sahu[2], Rakesh V Radhakrishnan[3]

Orbitofacial neurofibromatosis type 1 (OFNF) is a rare and unique variant of neurofibromatosis type 1. This case report aims to highlight the clinical observations and surgical management of a patient with a large atypical cavernous hemangioma, which was masquerading as a plexiform orbitofacial neurofibroma. A 35-year-old female presented with a huge orbitofacial mass, which was clinically and radiologically diagnosed as an orbitofacial neurofibroma involving the right eye and face. A detailed history and physical examination were performed, and a plastic surgery consultation was sought. The patient was diagnosed with OFNF. After conducting an orbit and brain MRI, along with other routine investigations, surgery was planned. The patient underwent right eye exenteration with an ipsilateral pedicled temporoparietal fascia flap covered with a split-thickness skin graft. Histopathology revealed the mass to be a cavernous hemangioma. In such a rare condition, an incisional biopsy may guide further definitive surgical treatment, ensuring that the final outcome involves minimum possible disfiguration and has the least impact on the quality of life of the patient.

Key words: Cavernous hemangioma, exenteration, masquerading, orbitofacial mass, pedicled temporoparietal fascia flap, plexiform orbitofacial neurofibroma

Neurofibromatosis type 1 (NF1), also known as von Recklinghausen disease, is not very rare, occurring in approximately 1 in 3000 live births. It is an autosomal dominant neurocutaneous disorder with diverse clinical manifestations.[1] Most commonly, NF1 presents as benign tumors that grow slowly; however, these tumors can lead to significant functional and cosmetic impairment. Malignant transformation occurs in approximately 5%–10% of patients with NFI, particularly in the subtype known as plexiform neurofibromas.[2]

Orbitofacial neurofibromatosis type 1 (OFNF) is a rare and unique variant of NF1. OFNF affects approximately 1%–22% of patients and is characterized by progressive tumors that cause disfigurement in the eyelid, eyebrow, temple, face, and orbits.[1] Tumors involving the orbit in NF1 are often more aggressive and invasive compared to those found in other parts of the body, with a higher recurrence rate after surgical excision. In this report, we present a case of a 35-year-old female with a massive orbitofacial lesion mimicking plexiform OFNF, which resulted in significant visual impairment and severe cosmetic effects.

Cavernous hemangioma in infants typically resolves spontaneously and may have a dramatic course.[3] Conversely, cavernous hemangiomas that present in adulthood, although they may originate in childhood, tend to remain stable in the early stages of the disease. However, they can cause significant deformity and distress.[3] It is very rare to find a cavernous hemangioma that mimics a plexiform orbitofacial neurofibroma, involving the eye, orbit, and one side of the face. Due to the limited literature available on this topic, we herein aim to report a case of a large cavernous hemangioma affecting one side of the face and the orbit, which presented as a plexiform neurofibroma on imaging.

Case Report

A 35-year-old female presented with swelling involving her right eye since she was 6 months old. The swelling gradually increased in size, with rapid growth occurring in the past year. Later, it became associated with pain and discharge, along with a loss of vision over the past year. The pain radiated to the neck. Ocular examination revealed a 10 cm × 6 cm lesion involving the right upper and lower eyelids, extending to the right supraorbital margin, and displacing the adnexa and eyeball inferolaterally with abaxial proptosis [Fig. 1]. The face was asymmetric, with diffuse soft tissue involvement on the right side, along with total ophthalmoplegia in the right eye. The entire orbit was involved, and the mass was soft in consistency, nontender, and nontranslucent, with no bruit or pulsation felt. There was a serosanguinous discharge from the right eye, along with conjunctival chemosis. The cornea was clear, with a mid-dilated and fixed pupil, and there was no perception of light. Fundus examination revealed optic atrophy in the right eye, with a normal left eye. No swelling was found elsewhere in the body, and other systems were grossly normal. Her past medical history was unremarkable, and her immunizations were completed according to her age and the government schedule. There was no family history of such an illness. Routine laboratory investigations were all normal. MRI revealed a huge mass measuring approximately 8.9 × 6.8 × 7.4 cm. The mass was heterogeneously hypointense on T1 and heterogeneously hyperintense on T2 images. It was a right intra- and extra-conal lesion, likely a plexiform neurofibroma [Fig. 2]. The diagnosis was clinically more in favor of orbitofacial plexiform neurofibroma, with a possible differential of lymphangioma. The patient was planned for debulking and exenteration with reconstruction after the patient and her family were thoroughly educated and counseled. Intraoperatively, after debulking and exenteration, no abnormalities were observed in the orbital bone. The lateral wall, roof, and medial wall of the orbit were denuded of periosteum. The temporoparietal fascia was harvested, based on the superficial temporal artery and vein (temporal branch) [Fig. 3]. The skin over the zygomatic-temporal region was undermined to create a tunnel for the TP fascia to pass into the orbit. The fascia was spread over the exposed bony socket, fixed at the margins, and secured with an anchorage suture at the apex of the orbit [Supplementary Fig.]. A split-thickness skin graft was harvested from the thigh and applied over the fascia and residual soft tissue. Histopathology sections revealed numerous large, variable-sized spaces filled with blood and lined by endothelial cells [Fig. 4. H and E stain 200x], suggesting a daignosis of cavernous hemangioma. A one year follow-up indicated no recurrence. The patient is well and continues with her daily activities. Informed written consent was obtained from the patient for publication of the case, including clinical images.

Discussion

Managing large hemifacial plexiform neurofibromas poses a significant challenge and requires a multidisciplinary approach along with careful preoperative planning. The primary aim is

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DOI: 10.4103/IJO.IJO_3190_23

Department of Ophthalmology, [2]Burns and Plastic Surgery, and [3]College of Nursing, All India Institute of Medical Sciences, [1]Department of Plastic and Reconstructive Surgery, Kalinga Institute of Medical Sciences, Bhubaneswar, Odisha, India

Correspondence to: Dr. Sandip K Sahu, Department of Ophthalmology, All India Institute of Medical Sciences, Bhubaneshwar - 751 019, Odisha, India. E-mail: drsandipsahu@yahoo.com

Received: 06-Dec-2023 Revision: 22-Jun-2024
Accepted: 02-Sep-2024 Published: 01-Nov-2024

Cite this article as: Sahu SK, Rout SK, Sahu RK, Radhakrishnan RV. Atypical cavernous hemangioma clinically masquerading as a plexiform Orbitofacial neurofibromatosis: A case report. Indian J Ophthalmol Case Rep 2024;4:807-10.

to enhance both function and aesthetics. Patients with facial disfigurement may experience social withdrawal and suffer from negative psychological effects. The weight of the tumor can lead to displacement of the eye and, in severe cases, result in loss of vision and disfigurement. Surgery can positively impact multiple aspects of a patient's life.

Conversely, cavernous hemangioma is a common vascular lesion in the orbit among adults. Over 80% of these lesions are found within the intraconal compartment, with the lateral side being the most common location.[4] They are more prevalent in women (60%–70%) during their forties and fifties. Extraconal hemangiomas, especially in atypical areas like the nose and paranasal sinuses, are rare and difficult to diagnose.[5,6] Limited literature is available regarding extraconal orbital hemangiomas situated on the inferior rim of the orbit.[7] To the best of our knowledge, no literature exists on cases involving both the extra- and intraconal regions of the entire orbit. However, intraconal hemangiomas tend to grow slowly, with a gradual onset of symptoms such as proptosis or mass effect. The diagnosis is typically supported by MRI images. However, in our case, the mass appeared hypointense on T1 and hyperintense on T2 images, which is unusual. This mass was located within both the intra- and extraconal areas of the right globe, suggesting a likely plexiform neurofibroma. In our case, the mass had a significant effect on the orbit and face, resembling a plexiform orbitofacial neurofibroma. Giant orbitofacial neurofibromas are typically treated surgically, but complete resection is often challenging due to the extensive growth of the tumor, and there is a risk of developing malignant changes.[7]

Histopathology of cavernous hemangiomas reveals multiple large vascular channels covered by endothelial cells and a substantial amount of stroma.[8] The vascular lumen is filled with blood, and there may be areas of thrombosis, indicating slow flow or stasis. The endothelial cells appear as mature vascular elements, and the stromal structure demonstrates increased cellularity or hyperplastic elements associated with neovascular activity.[8] Conservative management is typically recommended for cavernous hemangiomas, while surgical

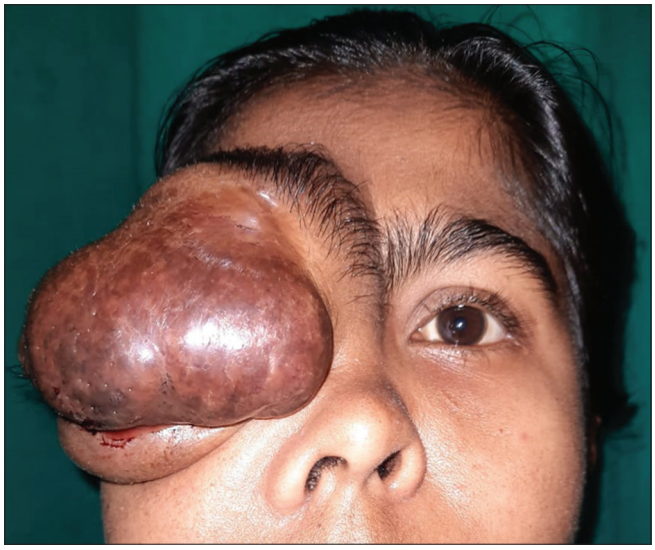

Figure 1: Preoperative image of the patient showing a 10 cm × 6 cm lesion involving the right eye with abaxial proptosis

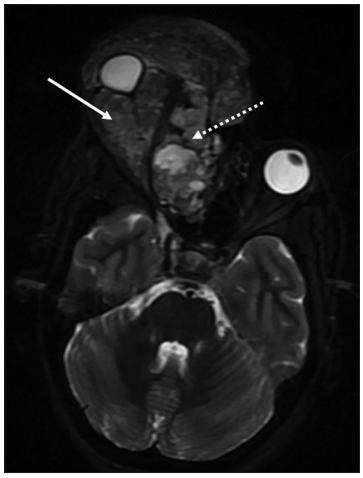

Figure 2: Axial T2WI of the orbit showing a heterogeneously hyperintense mass in the right orbit involving both intraconal (arrow) and extraconal spaces (arrowhead) with protrusion of the globe

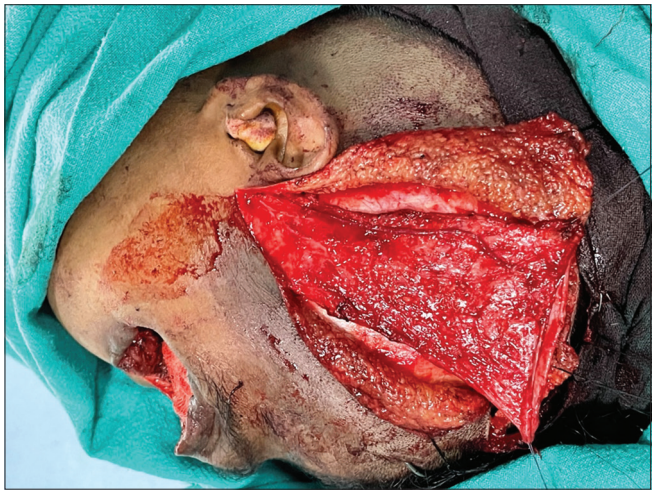

Figure 3: Image depicting the harvested temporoparietal fascia based on the superficial temporal artery and vein

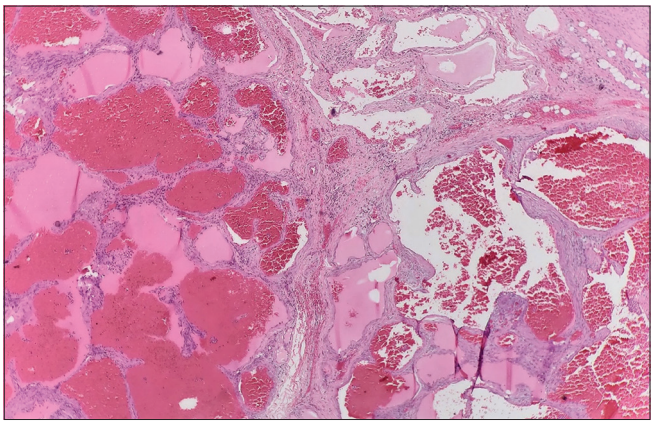

Figure 4: Sections show numerous variable-sized, thin-walled blood vessels filled with blood. Vessels are lined by endothelial cells. H and E stain, 200×

excision is reserved for cases that cause severe proptosis or compression of the optic nerve,[9] such as in our case of unilateral hemangioma.

Both cavernous hemangioma and OFNF are orbital tumors. Due to the different treatment strategies and intraoperative courses of these tumors, it is crucial to differentiate between them at the start of treatment. Recently, the use of artificial intelligence (AI) in medicine has demonstrated classification accuracy comparable to that of physicians in diagnosing various diseases.[10] Developing an AI framework that utilizes MRI image sets and clinical data from multiple centers automate the process of differentiating between cavernous hemangioma and OFNF with high levels of accuracy, sensitivity, and specificity.

Conclusion

Although rare, a giant cavernous haemangioma may clinically and radiologically mimic plexiform neurofibroma, especially in the orbitofacial region. An incisional biopsy or AI may enable physicians to arrive at an accurate diagnosis. Both cavernous haemangioma and plexiform neurofibroma are ill-defined and may be subjected to incomplete excision. Regular follow-up is necessary for plexiform neurofibroma due to the risk of malignant transformation. Surgical excision should be carefully balanced to minimize the recurrence and achieve optimal aesthetics.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Financial support and sponsorship: Nil.

Conflicts of interest: There are no conflicts of interest.

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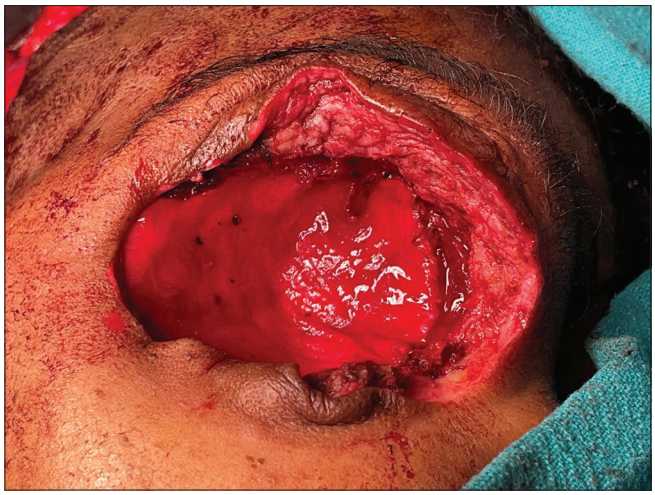
DOI: 10.4103/IJO.IJO_3190_23



Supplementary Figure: The image depicting the fascia that was spread over the exposed bony socket and fixed at the margins and one anchorage suture was applied at the apex of the orbit