CASE REPORT

Isolated Cervical Plexiform Neurofibroma Mimicking Hemangioma: A Rare Case Report

Ramya Rajpurohit[1], Deepak Verma[2], Naveen Sharma[3], Shubham Bhatnagar[4]

Received on: 24 January 2025; *Accepted on*: 28 July 2025; *Published on*: XX XXXX XXXX

Abstract

Neurofibromas are benign soft tissue tumors affecting young adults between the ages of 20–30 years and account for almost 5% of all soft tissue tumors. Histopathologically, they are classified as local, diffuse, and plexiform. Of these, plexiform neurofibromas (PNF) are benign tumors of the peripheral nerve that predominantly develop in patients with neurofibromatosis type 1 (NF1), but isolated cases of plexiform neurofibroma without any association with NF1 have also been reported in the literature. The infrequency of isolated PNF and its clinicoradiological presentation further makes it a rare and a diagnostic challenge. Here, we report the case of a 6-year-old boy who presented with gradually progressive left neck swelling which was provisionally diagnosed as a case of a vascular malformation in the preoperative period. However, it subsequently turned out to be a plexiform neurofibroma on final histopathological examination. Hence this case is being reported here for its rarity and diagnostic challenge.

Keywords: Case report, Hemangioma, Neurofibromatosis, Plexiform neurofibroma.

Otorhinolaryngology Clinics: An International Journal (2025): 10.5005/jp-journals-10003-1686

Introduction

Plexiform neurofibromas (PNF) are benign tumors originating from nerve sheath cells of subcutaneous or visceral peripheral nerves.[1] Noted predominantly in patients with NF1, it is caused by a defect of the tumor suppressor gene *NF1* on 17q11.2, including single base substitutions, splice site mutations, or single base deletions, resulting in abnormal neurofibromin. The *RAS* gene is downregulated by neurofibromin, mutation of which ensues to multiple neurofibroma formation.[2] Approximately, a third of neurofibromatosis type 1 (NF1) patients suffer from PNF, which is often present at birth.[3] Though frequently regarded as pathognomonic of NF1, isolated PNF has also been documented.[4,5] These slow-growing tumors have variable symptomatology depending on their topography.[6] In the paucity of a clinical picture of NF1, the diagnosis of an isolated plexiform neurofibroma is typically anatomopathological. The clinical presentation may not be specific, likening a vascular malformation, or a benign/malignant tumor of skin or connective tissue.[7,8]

Case Description

Herein is the rare case of a 6-year-old boy who was brought to the otorhinolaryngology clinic with the complaint of a painless soft tissue swelling involving the left side of the neck. Swelling was first noted by the parents a month ago. Initially, it was pea-sized, but gradually progressed to the presenting size of approximately 5 × 3 cm corresponding to level II and III lymph node levels. Skin over the swelling was normal with no discoloration, scar, or sinus. The lesion was mildly tender, diffuse with ill-defined margins, soft in consistency, nonpulsatile, and had a characteristic bag of worms appearance on palpation. Thus, a provisional diagnosis of a vascular malformation was considered (Fig. 1).

The patient underwent ultrasonography, which revealed multiple anechoic to hyperechoic areas in the substance of the left sternocleidomastoid muscle consistent with the findings of

[1–4]Department of Otorhinolaryngology, Bhagat Phool Singh Government Medical College for Women, Sonipat, Haryana, India

Corresponding Author: Ramya Rajpurohit, Department of Otorhinolaryngology, Bhagat Phool Singh Government Medical College for Women, Sonipat, Haryana, India, Phone: +91 8890259041, e-mail: rajpurohit.ramya@gmail.com

How to cite this article: Rajpurohit R, Verma D, Sharma N, *et al*. Isolated Cervical Plexiform Neurofibroma Mimicking Hemangioma: A Rare Case Report. Int J Otorhinolaryngol Clin 2025;x(x):xx–xx.

Source of support: Nil

Conflict of interest: None

Patient consent statement: A written informed consent was obtained from the patient for the publication of details, which can include photographs and/or videos and/or case history to be published in any printed/online journals.

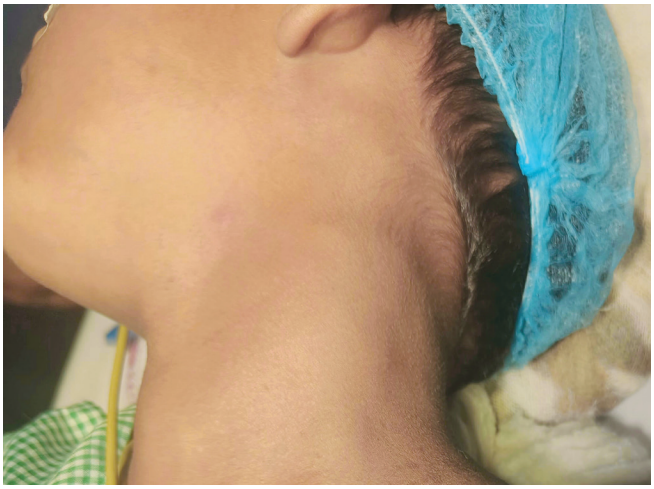
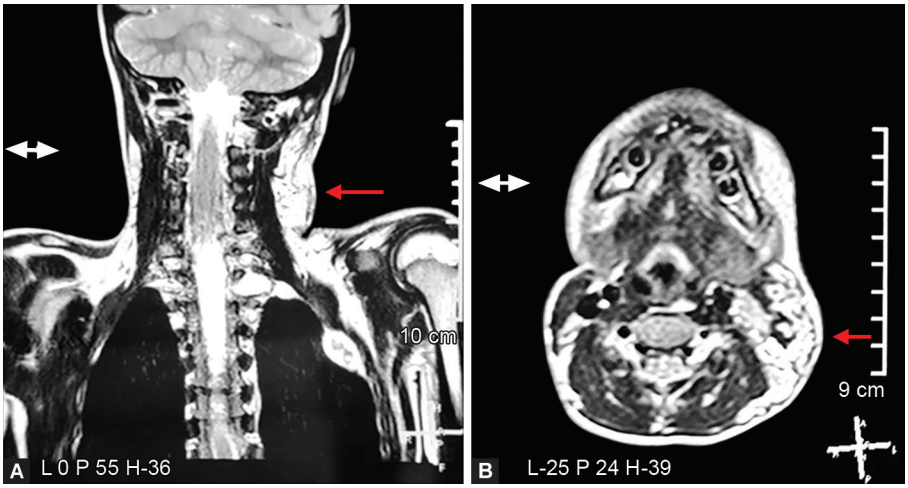


Fig. 1: Preoperative left cervical swelling



Figs 2A and B: CEMRI neck (A) Coronal section; (B) Axial section showing an altered signal intensity lesion with post-contrast heterogeneous enhancement with hypointense foci in skin, subcutaneous plane in the left cervical region (red arrow)

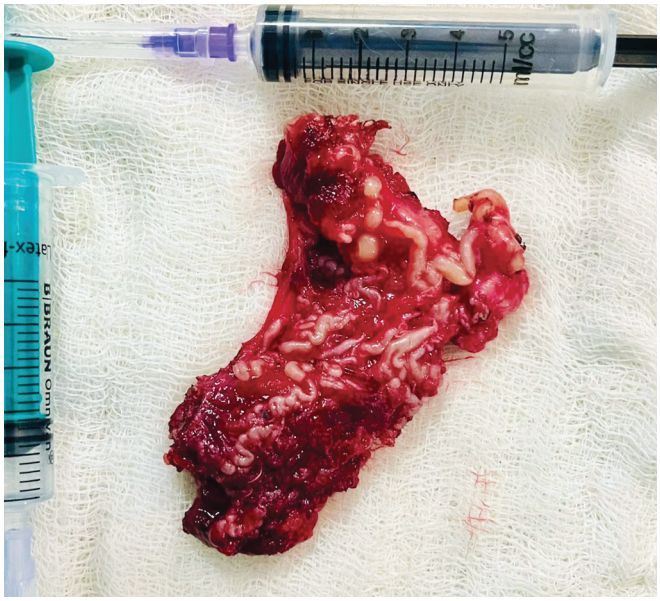


Fig. 3: Excised specimen

intramuscular hemangioma. CE-MRI was suggestive of a 51 × 36 mm altered signal intensity lesion with postcontrast heterogeneous enhancement with multiple T2 hypointense foci in the skin and subcutaneous plane in the left cervical region, further confirming the provisional diagnosis of a vascular malformation, most likely hemangioma. Multiple enlarged necrotic lymph nodes in left level II and V were also noticed, the largest measuring 20 × 16 mm (Fig. 2). CT-angiography revealed a lobulated marginated lesion in the subcutaneous plane in the left cervical region, with minimal peripheral enhancement of arterial phase and peripheral dilated tortuous vessels, likely low-flow vascular malformation. However, no obvious feeder vessel was detected.

Surgical excision of the cervical swelling was done under general anesthesia, along with excision of the level II lymph node (Fig. 3). To our surprise, histopathological examination revealed an expansile proliferation which demonstrated tortuous bundles of bland cells with elongated and wavy nuclei in background of myxoid changes, without any evidence of atypia, necrosis or mitosis and a histopathological diagnosis of plexiform neurofibroma was made. The excised lymph node revealed inflammatory cells, which were in favor of reactive nodal hyperplasia.

Discussion

The *NF1* gene on the long arm of chromosome 17 encodes a 2818 amino acid protein product, neurofibromin, which is a small protein involved in promoting cell growth and differentiation. Neurofibromin functionally converts Ras to its inactive form, thus switching off cell growth. When a mutation in NF1 causes loss of normal neurofibromin activity, Ras remains active, and dysregulated cell growth and tumor formation can occur.[2] Plexiform neurofibromas are benign peripheral nerve sheath tumors that can be seen in either type of neurofibromatosis. They can be deep or superficial in location or a combination of the two. However, isolated occurrences of neurofibromas, in the absence of the classical clinical picture of NF-1, have also been reported. These have been postulated to arise from smaller dermal nerves in contrast to syndrome-associated PNF.[9,5,10]

Plexiform neurofibromas may cause symptoms ranging from cosmetic deformity to pain. It has a typical bag-of-worm appearance on palpation, as appreciated in the present patient.[9,5] As observed in this case, these lesions may seldom demonstrate a vascular nature, both clinically and radiologically.[11] Similar to the present case, studies have also concluded that imaging modalities may show overlapping features between neurofibromas and vascular malformations. Ultrasonography, which is the initial investigative modality, may show hyperechoic regions fenestrated by blood vessels, which are represented by hypoechoic foci, with or without infiltration into adjacent structures.[4] On MRI, the central dark focus, termed the target sign, is present in deep PNF on T2-weighted or STIR images, and can also mimic phleboliths, common in venous malformation. Superficial neurofibromas may have diffuse rather than fascicular or nodular morphology, and the nodules and fascicles are smaller. Their signal characteristics are usually homogeneous or heterogeneous, sans the characteristic target sign, which is a feature of their deeper counterparts.[11,12]

Histology may demonstrate an unencapsulated or circumscribed lesion, with neoformed cystic tumor proliferation and

spindle-shaped cells with corrugated nuclei, in the background of abundant collagen. Myxoid and degenerative regions may also be seen. S-100 protein, CD-34 maybe positive but is of limited value. Malignant transformation is usually not encountered.[5,9]

Management of isolated PNF is surgical resection. If neurofibromas cannot be easily separated from the underlying normal nerve, their complete excision may warrant the sacrifice of the nerve. This is preferred for superficial PNF and avoided for their deep counterparts due to the imminent risk of neurological deficit. In such cases, debulking or conservative management can be done. Risk of recurrence depends on distinct variables of patient age, tumor location, and extent of resection. Hence, it is prudent to adopt a tailor-made management plan on an individual basis. Malignant transformation of an isolated plexiform neurofibroma is a rare occurrence.[13]

Conclusion

To conclude, an isolated plexiform neurofibroma is a rare entity if not associated with neurofibromatosis. Furthermore, its equivocal clinical and imaging appearance may pose a diagnostic dilemma and may point towards a vascular malformation. Hence, knowledge of this infrequent entity with its characteristic clinical and imaging characteristics would lead to its timely diagnosis and appropriate subsequent management.

Declarations

Ethics Approval and Consent to Participate

This study was conducted in accordance with the ethical principles outlined in the Declaration of Helsinki and its amendments. The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient and guardian have given consent for their image and other clinical information to be reported in the journal. The patient and guardian understand that their name and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Written informed consent and necessary approval and consent to participate in written format were duly obtained from all subjects/participants. Care has been taken to preserve the identity of the patient.

Availability of Data and Material

Data sharing is not applicable to this article as no datasets were generated or analyzed during the current study.

Authors' Contributions

All authors discussed the results and contributed to the final manuscript.

References

1. Kleihues P, Cavenee WK. WHO classification of tumours. Pathology & genetics. Tumors of the Nervous System. Lyon, France: IARCpress, 2000. DOI: 10.1002/1096-9896(200102)193:2<276::AID-PATH765>3.0.CO;2-Q.
2. Jeong SY, Park SJ, Kim HJ. The spectrum of *NF1* mutations in Korean patients with neurofibromatosis type 1. J Kor Med Sci 2006;21(1):107. DOI: 10.3346/jkms.2006.21.1.107.
3. Rosenbaum T, Wimmer K. Neurofibromatosis type 1 (NF1) and associated tumors. Klinische Pädiatrie 2014;226(06/07):309–315. DOI: 10.1055/s-0034-1382021.
4. Siddiqui S, Khan ZA, Wahab S, et al. A rare case of diffuse neurofibroma masquerading as a hemangioma: A radiological perspective. J Ultrasound 2017;20:343–346. DOI: 10.1007/s40477-017-0268-z.
5. Ahmedou AB, Amine MM, Youssef O, et al. A rare cause of cervical swelling: Solitary plexiform neurofibroma. Ann Med Surg 2021;64:102225. DOI: 10.1016/j.amsu.2021.102225.
6. Bouimetarhan L, Bellamlih H, En-Nafaa I, et al. Plexiform cervical neurofibroma: About a case. Pan Afr Med J 2018;30:41. DOI: 10.11604/pamj.2018.30.41.14446.
7. O'Keefe P, Reid J, Morrison S, et al. Unexpected diagnosis of superficial neurofibroma in a lesion with imaging features of a vascular malformation. Pediatr Radiol 2005;35:1250–1253. DOI: 10.1007/s00247-005-1565-9.
8. Ward BA, Gutmann DH. Neurofibromatosis 1: From lab bench to clinic. Pediatr Neurol 2005;32(4):221–228. DOI: 10.1016/j.pediatrneurol.2004.11.002.
9. Ho JD, Ho JA, Ruddock R, et al. Isolated, nonsyndromic mucocutaneous plexiform neurofibromas: A systematic review of the clinicopathologic features. Am J Dermatopathol 2022;44(12):904–912. DOI: 10.1097/DAD.0000000000002322.
10. Stefano PC, Apa SN, Lanoël AM, et al. Isolated plexiform neurofibroma mimicking a vascular lesion. An Bras Dermatol 2016;91:240–242. DOI: 10.1590/abd1806-4841.20164300.
11. Anand R, Yadav DS, Yadav V, et al, Bhatia D. Plexiform neurofibroma in a 16-year-old girl. Radiol Case Rep 2012;7(3):708. DOI: 10.2484/rcr.v7i3.708.
12. Arnold R, Chaudry G. Diagnostic imaging of vascular anomalies. Clin Plast Surg 2011;38(1):21–29.
13. Atallah I, Gervasoni J, Gay E, Righini CA. A rare case study of a retropharyngeal neurofibroma and a brief literature review. Eur Ann Otorhinolaryngol Head Neck Dis 2016;133(1):47–50. DOI: 10.1016/j.anorl.2015.11.003.