International Journal of Innovative Research in Medical Science (IJIRMS)
Volume 09, Issue 05, May 2024,
https://doi.org/10.23958/ijirms/vol09-i05/1891

Case Report

Malignant Peripheral Nerve Sheath Tumour of Left Vagus: A Rare Presentation in Neurofibromatosis-1

Dr Dhamodhara Kannan Shivarajan [1], Dr Siddharth P. Dubhashi *[2], Dr Bhupendra Mehra [3], Dr Divish Saxena [4],

[1]Junior Resident, Department of General Surgery, All India Institute of Medical Sciences, Nagpur, India.
[2]Professor and Head, Department of General Surgery, All India Institute of Medical Sciences, Nagpur, India.
[3]Additional Professor, Department of General Surgery, All India Institute of Medical Sciences, Nagpur, India.
[4]Associate Professor, Department of General Surgery, All India Institute of Medical Sciences, Nagpur, India.

*Corresponding author: Dr Siddharth P. Dubhashi; *spdubhashi@aiimsnagpur.edu.in*

Received 19 April 2024; Accepted 11 May 2024; Published 16 May 2024

Abstract

Background: Malignant Peripheral Nerve Sheath Tumour (MPNST) is a rare but aggressive type of soft tissue tumor. In cases associated with neurofibromatosis, where they often develop from existing plexiform neurofibromas, prognosis is poorer compared to sporadic cases. MPNST originating from a cranial nerve is extremely uncommon, requiring systematic diagnosis and surgical management based on thorough history, clinical examination, and lab tests. ***Case report:*** Our case involves a young adult male who presented with a 10 x 8 cm neck mass in the left anterior triangle, alongside other neurofibromatosis features like cafe-au-lait spots and Lisch nodules. MRI and CECT revealed a well-defined mass from the left Vagus nerve, adjacent to the left carotid artery and jugular vein. Biopsy confirmed neurofibroma, and the mass was surgically excised with negative margins, revealing MPNST upon histopathology. ***Conclusion:*** The uncommon occurrence of MPNST originating from a cranial nerve, along with its potential for local invasion and distant spread, underscores the importance of early diagnosis and prompt surgical intervention.

Keywords: ***Neurofibromatosis; Malignant peripheral nerve sheath tumor; Vagus.***

Introduction

Malignant Peripheral Nerve Sheath Tumour (MPNST) is a highly aggressive tumour accounting for 5-10% of all soft tissue tumours [1]. In association with neurofibromatosis, these tumors have higher chances of malignant transformation from pre-existing plexiform neurofibromas,[2] thereby carrying a worst prognosis, as compared to sporadic origin. MPNST from a cranial nerve is an exceedingly rare phenomenon [3]. The diagnosis and surgical management of such an aggressive tumour requires a systematic and anatomical approach, based on careful history, clinical examination and laboratory investigations.

Case Report

A 20-year-old boy presented with a slowly increasing swelling on left side of neck for 1 year. (**Fig 1**), associated with change in voice and cough. There were no comorbidities. On examination, there was a swelling of 10x6 cm noted over left carotid triangle of neck with scar of previous biopsy procedure, along with other features of neurofibromatosis like – café au lait spots and Lisch nodules on slit lamp examination. Magnetic Resonance Imaging of neck revealed abnormal signal intensity on left side of neck along posterolateral aspect of carotid sheath. Contrast-enhanced Computed Tomography = neck (**Fig 2**) showed a well-defined encapsulated heterogenous soft tissue mass at level II, III, IVa on left side of neck, compressing left Internal Jugular vein and displacing carotids anteriorly. Core cut biopsy of the lesion revealed neurofibroma. There was no evidence of metastases.

The lump was excised in-toto. Intra-operatively tumor mass was found to be abutting Left carotid and left Internal Jugular vein - arising from left Vagus. Histopathological examination revealed a malignant peripheral nerve sheath tumor (MPNST) (Fig 3) of left vagus with negative margins. Post operative course was uneventful. Patient was given adjuvant radiotherapy. The follow-up period of 2 years has been uneventful.

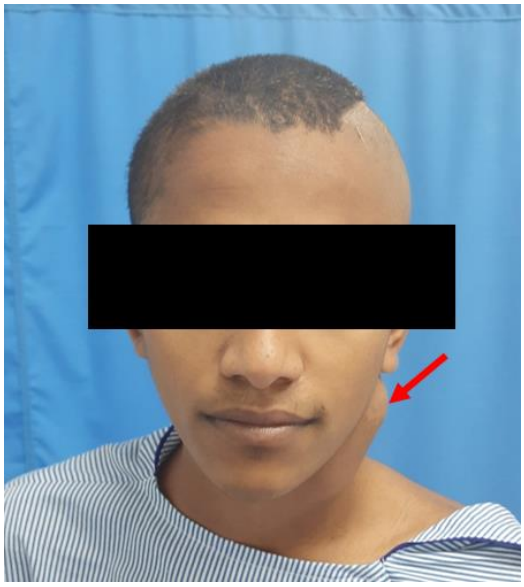

Fig 1: Swelling over left side of neck

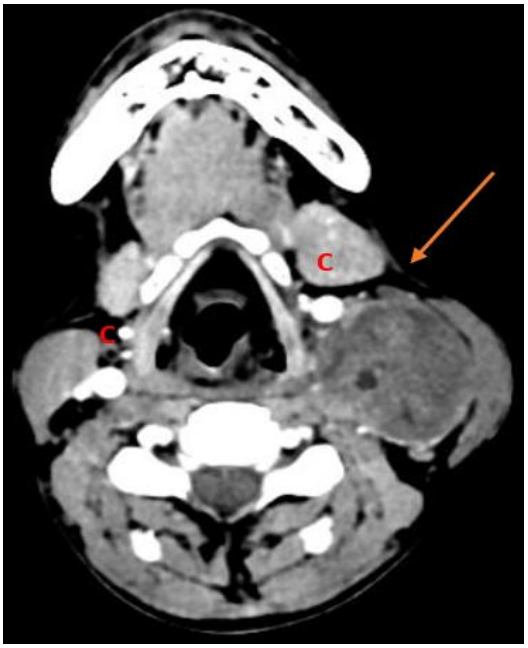

Fig 2: Axial section Contrast enhanced CT Head and Neck: Well-defined encapsulated soft tissue density mass. Central calcific foci denoted by orange arrow and Carotids denoted by (C) are displaced anteriorly.

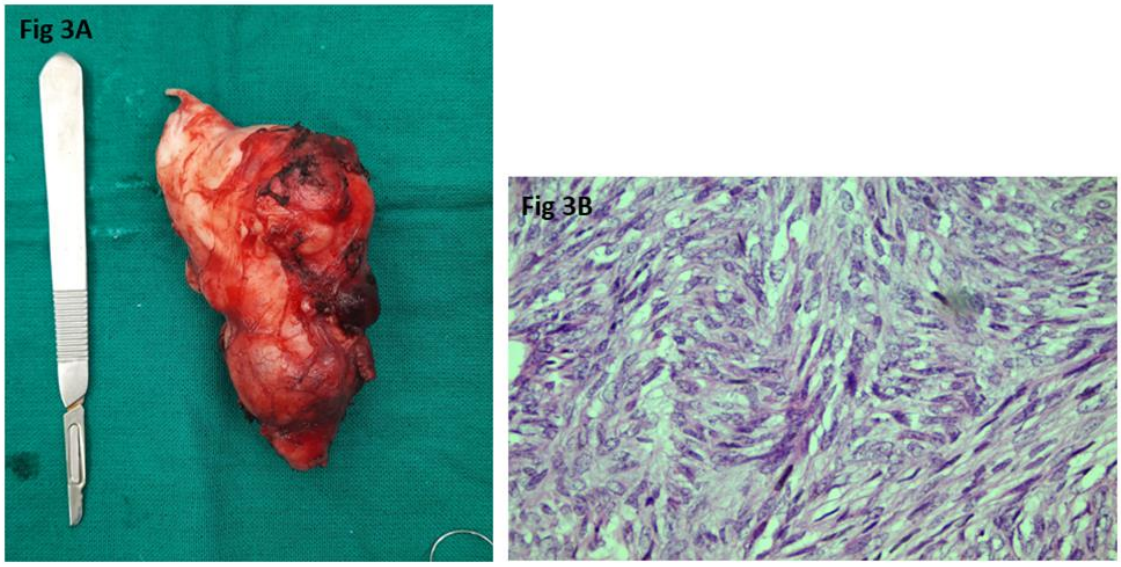

Fig 3: 3A: Gross Specimen showing irregular bosselated whitish mass with cut surface showing focal necrotic and haemorrhagic areas. 3B: Histopathological examination showing Herring bone pattern with areas of hyalinisation in High power (40X)

Discussion

Malignant Peripheral Nerve Sheath Tumors (MPNST) are extremely rare soft tissue tumor (5- 10% incidence among all soft tissue tumors) arising from peripheral nerves, most commonly affecting trunk and extremities.[1] MPNST was categorised as a soft tissue sarcoma by WHO in 2013 [4]. Neurofibromatosis - Von Recklinghausen's disease patients are predisposed for malignant transformation of pre-existing neurofibromas [5]. Radiation exposure is also a recognised risk factor. MPNSTs usually arise from medium to large size nerves. MPNSTs can also arise from cranial nerves. High index of suspicion of MPNST should be there when the patient has features of neurofibromatosis or when the tumour is obviously arising within the anatomical compartment of a major nerve or in continuity with neurofibroma. Patients usually present with rapidly enlarging mass, causing pain or local neurological symptoms [6]. 50% of cases present with lung metastasis. This was not seen in our case.

Lateral neck mass opens up for a wide varied number of differentials like lipoma / salivary gland tumors / vascular tumors / carotid body tumors / branchial cleft cyst, posing a diagnostic dilemma. Imaging findings may help in identification of origin site. Magnetic Resonance Imaging is superior in view of better soft tissue delineation and indication of neuro vascular involvement. MPNST on histopathology, is characterised by differentiated Schwann cells, fibroblasts, vascular cells and mast cells. The spindle cells are arranged in fascicular pattern. They should also be distinguished from malignant granular cell tumor, also of Schwannian origin. Cell necrosis, haemorrhage and mitosis are common [6].

Mainstay of treatment is surgical excision, with an aim of margin-negative resection. Intra-operatively, dissection of the lesion from major nerves and vessels is a challenge and appropriate measures for vascular and nerve repair to be prepared beforehand. Lymph node involvement in such tumors is again a rare phenomenon, and if found intra-operatively, to be sent for histopathological examination. Adjuvant radiotherapy may be required [7]. Chemotherapy is an option for unresectable / metastatic MPNST. Trials for targeted therapy have been conducted for EGFR inhibitor (Erlotinib) and MEK inhibitors (Mirdamitinb) and selective kinase inhibitors for BRAF V600 (Vemurafinib).

Survival rates are very low with MPNSTs. Various prognostic indicators for MPNST include – large size at presentation (>5cm), tumor grade, status of surgical margin and local recurrence [8]. Minovi *et al* has reported 5-year survival rates between 15% and 47% in the general population, with lower rates for patients with the NF1 mutation (23%) compared with others (47%) [9]. The reasons for these differences are related to a tendency toward larger tumors, poorer differentiation, higher rates of metastases, and multifocality for these patients. Wu Y *et al* proposed that atypical neurofibromatosis (ANF) serves as an intermediate stage in the transformation of benign peripheral neurofibromas (PN) into malignant peripheral nerve sheath tumors (MPNST) [5]. The progression from PN to ANF is primarily influenced by the deletion of CDKN2A/B genes. Further advancement from ANF to MPNST may entail significant chromosomal rearrangements and frequent inactivation of the PRC2 gene.

Conclusion

MPNST are uncommon biologically aggressive lesions with poor prognosis, demonstrating propensity for rapid disease progression. Therapeutic options are limited. Complete surgical excision is the most effective modality. Evidence based protocols should provide a consensus for combination of chemo-radiation and targeted therapy for optimal outcome.

Acknowledgements

None

Conflict of Interests

We declare no conflict of interests.

Funding Statement

Not Applicable

References

[1] Gupta G, Maniker A. Malignant peripheral nerve sheath tumors. Neurosurg Focus. 2007;22(6):E12.

[2] Fasih S, Suppiyah S, Barron J, Barnett-Tapia C, Avery R, Dickson B, Ferguson P, Swallow C, Zadeh G, Gupta AA. Malignant transformation of plexiform neurofibroma to MPNST while on MEK inhibitor. Neurooncol Adv. 2021;3(1):1-2.

[3] Scheithauer, Bernd W. MD; Erdogan, Sibel MD; Rodriguez, Fausto J. MD; Burger, Peter C. MD; Woodruff, James M. MD; Kros, Johan M. MD; Gokden, Murat MD; Spinner, Robert J. MD. Malignant Peripheral Nerve Sheath Tumors of Cranial Nerves and Intracranial Contents: A Clinicopathologic Study of 17 Cases. The American Journal of Surgical Pathology March 2009; 33(3): 325-338.

[4] Dukatman BS, Scheithauer BW, Piepgras DG, Reiman HM, Ilstrup Dm. Malignant peripheral nerve sheath tumors. A clinico pathologic study of 120 cases. Cancer 1986; 57:2006-2021

[5] Wu Y, Cheng W, Zhang X, Li Z, Liu Y, Bai W. Neurofibromatosis type I-associated malignant peripheral nerve sheath tumors: a case report and literature review. Arch Med Sci. 2020;16(6):1476-1482.

[6] Faridh M, Demicco EG, Garcia R, Ahn L, Merola PR, Sioffi A, Maky RG. Malignant peripheral nerve sheath tumors. Oncologist 2014;19(2):193-201.

[7] Kahn J, Gillespie A, Tsokos M, Ondos J, Dombi E, Camphausen K, Widemann BC, Kaushal A. Radiation therapy in management of sporadic and neurofibromatosis type 1-associated malignant peripheral nerve sheath tumors. Front Oncol. 2014;4:324.

[8] Zou C, Smith KD, Liu J, *et al*. Clinical, Pathological and molecular variables predictive of malignant peripheral nerve sheath tumors outcome. Ann Surg 2009; 249: 1014-1022.

[9] Minovi A, Basten O, Hunter B, Draf W, Bockmühl U. Malignant peripheral nerve sheath tumors of the head and neck: management of 10 cases and literature review. Head Neck. 2007;29(5):439-45.