



Case Report:

Malignant peripheral nerve sheath tumor of the cervical vagus nerve in a neurofibromatosis type 1 patient - An unusual presentation

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Citation: Gupta A, Bansal S, Bhagat S, Bahl A. Malignant peripheral nerve sheath tumor of the cervical vagus nerve in a neurofibromatosis type 1 patient - An unusual presentation. *Online J Health Allied Scs.* 2010;9(1):10

URL: <http://www.ojhas.org/issue33/2010-1-10.htm>

Open Access Archives: <http://cogprints.org/view/subjects/OJHAS.html> and <http://openmed.nic.in/view/subjects/ojhas.html>

Submitted: Mar 14, 2010; Accepted: Apr 8, 2010; Published: Jul 30, 2010

Abstract:

Malignant peripheral nerve sheath tumors (MPNST'S) of the head and neck comprise 2% to 6% of head and neck sarcomas. These tumors may arise as sporadic variants or in patients with neurofibromatosis (NF). Development of these MPNST's is one of the serious complications of neurofibromatosis type 1(NF1). To our knowledge there are only two reported cases of MPNST's arising in the cervical vagal nerve, occurring in NF1 patients. We present here an NF1 patient who developed an MPNST of the cervical vagus nerve and presented only with a cervical swelling and hoarseness.

Key Words: Peripheral nerve sheath tumor, malignant, neurofibromatosis, vagus nerve

Introduction:

Malignant peripheral nerve sheath tumors (MPNST'S) of the head and neck comprise 2% to 6% of head and neck sarcomas. These tumors may arise as sporadic variants or in patients with neurofibromatosis (NF). The most common type of NF is NF type I (NF I) also known as von Recklinghausen's disease. This is an autosomal dominant disorder that demonstrates a marked variation in its expression. Development of these MPNST's is one of the serious complications of neurofibromatosis type 1(NF1). Their development is thought to be associated with both tumor suppressor gene mutations and deregulated growth factor signaling. MPNSTs of the cervical vagus nerve are extremely rare. To our knowledge there are only two reported cases of MPNST's arising in the cervical vagal nerve, occurring in NF1 patients.^{1,2}

The patients with NF should be closely monitored with a high index of suspicion towards rapidly enlarging, painful swell-

ings and associated with cough and hoarseness, as these are important features signifying malignant transformation. We present here an NF1 patient who developed an MPNST of the cervical vagus nerve and presented only with a cervical swelling and hoarseness.

Case Report:

A 34-year-old man with a known diagnosis of NF1 presented to the ENT outpatient department at PGIMER, Chandigarh with sudden hoarseness and swelling over the left upper neck for 2 years. He had a positive family history of NF1 affecting his paternal grandfather and father. Examination revealed a 8x9cm firm smooth, non tender swelling with restricted mobility present on the left side of neck posterior to mandibular angle. There was multiple small long- standing asymptomatic nodules present all over the body, clinically neurofibromas. There were multiple café au lait spots and freckles present over the shoulder. Laboratory results including routine blood counts, urinalysis, and chest-ray films were all unremarkable.

A complete paralysis of the left vocal cord was seen on flexible laryngoscopy examination. MRI showed a large well-defined heterogenous mass in the left carotid space that is mildly hyperintense to muscles on T2 weighted images. On T1 weighted images the lesion was isointense and showed intense enhancement in post contrast images (Fig. 1). It was displacing the common carotid and internal carotid artery anteriorly. Medially the wall of the oropharynx was deformed. Sternocleidomastoid muscle was displaced laterally and draped over the mass. No change of signal intensity or contrast enhancement was seen in the surrounding muscle to suggest invasion. Fine needle aspiration cytology was suggestive of nerve sheath tumor.

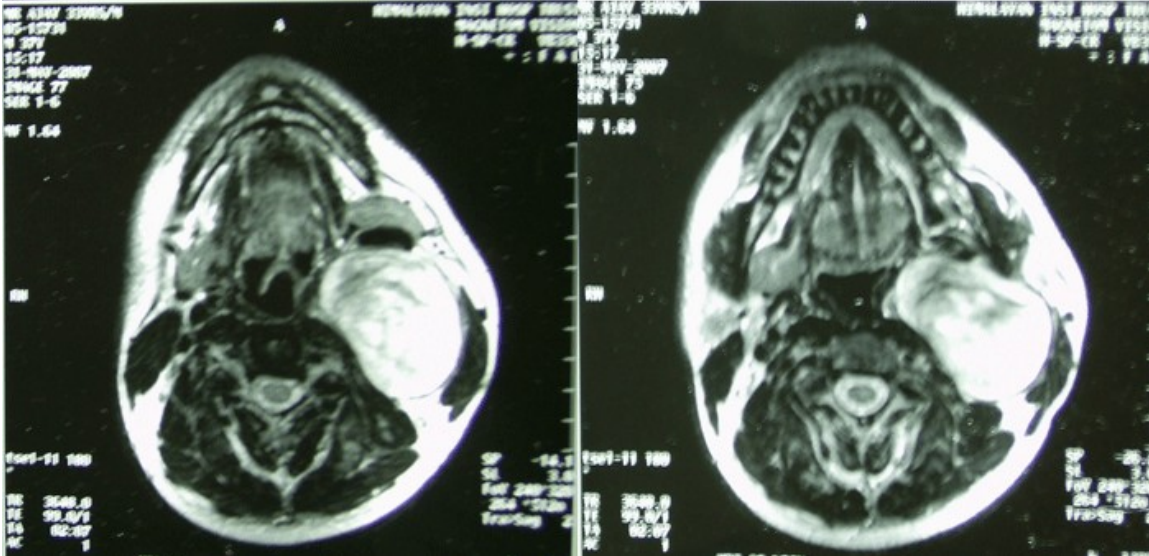


Fig. 1: MRI scans of the patient showing intense enhancement by the tumor

The patient underwent excision of the tumour by transcervical approach. A 10x8cm whitish, smooth, well-encapsulated tumor was seen arising from the vagus nerve and common carotid was found adherent to the capsule, which was freed. Tumor was excised completely along with the part of the vagus nerve engulfed by the tumor.

Histopathology showed overall features of malignant peripheral nerve sheath tumor composed of spindle shaped cells arranged in palisades and whorls. The tumor showed hyperchromatic nuclei and mitotic activity. Infiltration of the tumor cells into the surrounding soft tissue was noted (Fig. 2, 3, 4). Post-operatively the hoarseness of patient's voice worsened, possibly due to the loss of residual recurrent laryngeal nerve function. The patient underwent radiotherapy post operatively. The patient has been under regular follow up for the last 1 year and is free of any residual or recurrent tumor. A thyroplasty is now being planned for the patient.

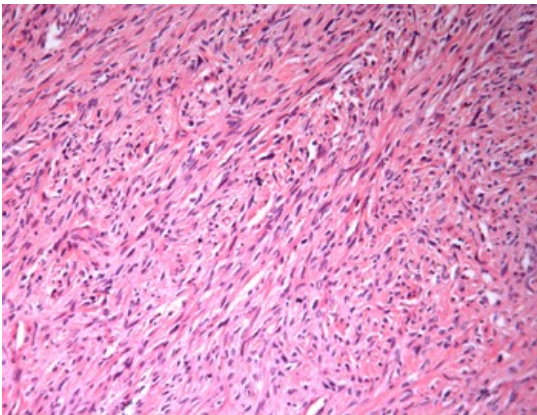


Fig. 2: Photomicrograph showing densely cellular areas alternating with less cellular areas. (Haematoxylin and eosin, x 100)

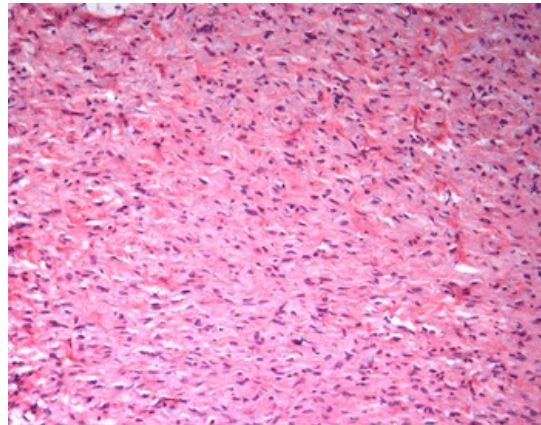


Fig. 3: Photomicrograph showing myxoid areas in which cells are having an irregular buckled shape characteristic of schwann cells. (Haematoxylin and eosin, x 100)

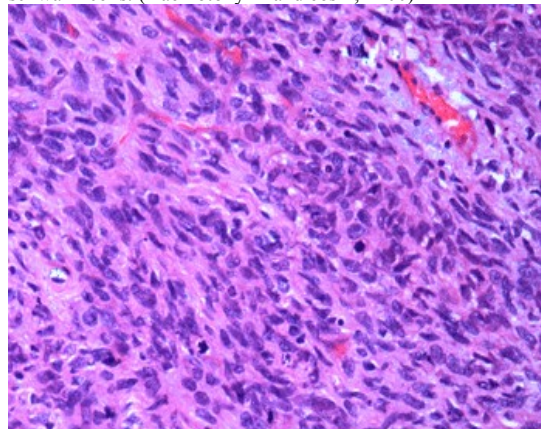


Fig. 4: Photomicrograph from cellular areas showing frequent atypical mitosis. (Haematoxylin and eosin, x 400)

Discussion:

Malignant peripheral nerve sheath tumors (MPNST'S) of the head and neck are rare tumors that may arise in a sporadic form or with in a pre-existing neurofibroma associated with neurofibromatosis. The limbs are the most common site for followed by the trunk, with head and neck being a very rare location.^{3,4} MPNST and neurogenic tumours of the vagus nerve are extremely rare. Gilmer-Hill et al⁵ reported a single case of neurofibroma and three cases of schwannomas of the cervical vagal nerve over a 31-year period. To our knowledge there are only four reported cases of MPNSTs arising in the cervical vagus nerve, two of which occurred in an NF1 patient.^{1,2}

The MPNSTs of NF1 may also be of low grade, although intermediate and high-grade tumours are more common.⁶ In our case a high-grade tumor was seen. MPNST commonly arises within a neurofibroma. The histological distinction between low- grade MPNSTs, neurofibroma with atypical features and neurofibroma is difficult. MPNSTs arising in a patient of NF1 are associated with poorer prognosis than in sporadic cases,⁷ with average five-year survival rates of just 16-21%.

Loree et al⁷ found the presence of neurofibromatosis and/or high-grade lesions was associated with statistically significant reduction in survival. The factors of age, head and neck site, and tumor size had no effect on survival. The decreased survival rate for MPNSTs in NF1 patients may be due to lower level of concern about new lesions leading to delayed presentation, rather than the cancer being inherently more aggressive. The lifetime risk of malignant transformation in NF1 patient has been reported as 1-2%.⁴ Radiation had been implicated as an etiological factor in the development of MPNST, with a latency period of 10 to 20 years.⁸ Other risk factors such as long- standing disease, particularly the presence of large number of plexiform neurofibromas from an early age, have been suggested.⁹

In a series of 34 NF1 patients who developed MPNST, none of the patient had a family member with MPNST.⁴ Monia et al., reported a case of MPNST of the cervical vagus nerve in a NF1 patient with a positive family history, as seen in our case. Certain familial mutations such as micro deletions of NF1 locus have been suggested to increase the risk of MPNST, although there has been no confirmatory study.⁹ Surgical resection of the tumor is the mainstay of therapy with in continuity resection of any involved neurovascular or bony structures.¹⁰ Local recurrence has been reported to occur in 40% to 50% of patients with MPNST.¹⁰ The use of radiotherapy has been advocated to improve the local control of disease.¹¹

The case discussed in this report highlights the importance of keeping a high index of suspicion towards a malignant transformation of these tumors though there were no classical fea-

tures of rapidly enlarging, painful swelling in association with cough and hoarseness especially in cases with NF 1. It is not clear whether the presence of MPNSTs in other NF1- affected family members predisposes the individual to a higher risk of malignant transformation.

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