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Case Report Open Access

Peripheral Nerve Sheath Tumour of Neck: A Rare Presentation a Case Report

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Introduction

Malignant peripheral nerve sheath tumour (MPNST) is a rare and very aggressive tumour of nerve cell origin associated with poor prognosis. Incidence of MPNST is 1 per 1, 00,000 populations and it constitutes between 3%-10% of all soft tissue sarcomas1-4. MPNST have been found to be associated with neurofibromatosis type 1 (associated with mutation in NF-1 gene) in 2%-29% cases [1-6]. Male and female are almost equally (53:47) involve [7]. These tumours often create diagnostic dilemmas because of non-specific clinical diagnostic criteria, histopathological resemblance with other spindle cell sarcomas like monophasic synovial sarcoma, leiomyosarcoma and fibrosarcoma [8]. Diagnosis is crucial because urgent intervention is necessary to prevent progression to inoperable stage. Here the authors present a case of MPNST of neck presenting as a small swelling, 2 cm in diameter, for a long period followed by rapid progression to a large size (7 cm 6 cm) causing impending obstruction of respiratory passage and encasement of great vessels of neck. The case is of particular interest because of its location, anatomical involvement, and diagnostic confusions by imaging and fine needle aspiration studies and perioperative management.

Case Presentation

A 19 years male patient presented with huge swelling over right side of neck (Figure 1). The swelling was small and very slowly increasing in size for last 14 years. There was a rapid increase in size of the swelling in previous 2 months. Patient also complained of pain over the swelling, head, upper back and right arm for previous one month. There was also small soft, fluctuant and transilluminant swelling in both upper and lower limbs for last 6 months. These symptoms were associated with decrease in appetite and weight loss. There was no history of difficulty in deglutition, difficulty in breathing, any abnormal respiratory sounds or blood in sputum. On examination there was a 6 8 cm swelling at the right side of neck almost occupying the entire right neck. The swelling was irregular, firm, no fixity to the skin. There was no venous engorgement over and above the swelling. Chest x-ray PA view revealed normal lung shadows. Plain and contrast enhanced CT scan of neck revealed a large, ill defined, non-enhancing hypodense lesion in the right side of neck extending from base of the skull to upper level of superior mediastinum occupying the right parapharyngeal and carotid space. The swelling was posterior and lateral to the paravertebral muscle displacing the right sternocleidomastoid muscle and medially extended upto the paravertebral space. There was an enhancing area seen within the lesion at the right submandibular region. Internal septations were seen in the lesion.

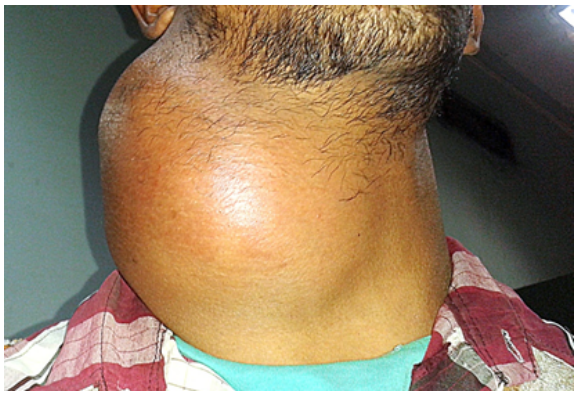

Figure 1: Patient presented with a large neck mass.

The larynx, pharynx and thyroid glands were displaced by the lesions. No abnormality was found in vessels of neck. The MRI scan revealed similar result. Internal lobulations and relations to the vessels were better delineated with MRI. The lesion was completely encasing the right internal and external carotid artery and partially encased the common carotid artery without any obliteration of the lumen. MRI also revealed presence of esophageal compression. Fine needle aspiration cytology revealed lymphoid tissue including small lymphocytes, plenty of large lymphoid cells and fair number of macrophages. No epithelioid cell, Langhans giant cell or Reed-Sternberg cell was detected. A provisional diagnosis of lymphovascular malignancy was considered from the imaging and fine needle aspiration studies. Debulking of the mass was planned for impending respiratory obstruction and patient was prepared for surgery. As the mass was encasing the great vessels and deformed the anatomy of upper aerodigestive tract, difficult airway was anticipated and awake nasal fibreoptic intubation with a wire reinforced endotracheal tube was performed under local anesthesia. Tranexamic acid was used to reduce perioperative bleeding. The surgical steps included an S shaped incision extending from 1 cm below pinna of right ear to right sternoclavicular joint (Figure 2).

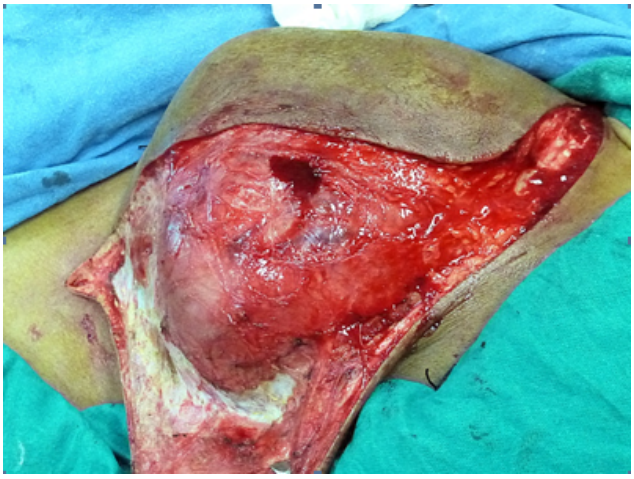

Figure 2: Shaped skin incision.

A large tumour mass was found extending from base of the skull to superior mediastinum encasing right internal and external jugular vein, right external carotid artery (Figure 3) and right brachial plexus.

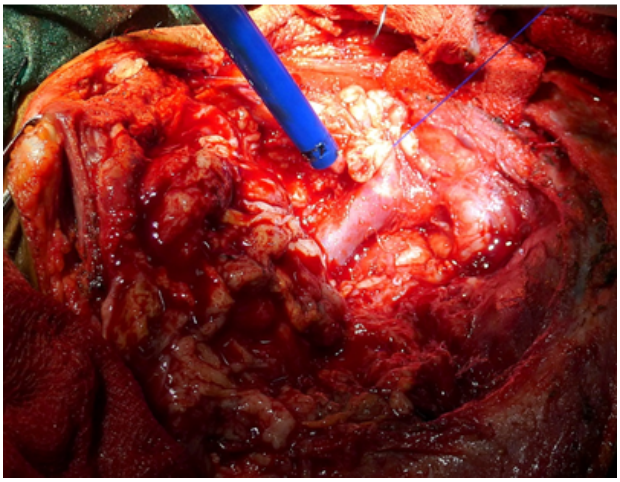

Figure 3: Tumour mass in relation to great vessels.

As the mass was inoperable debulking was done to relieve compression over trachea and other major structures. Hemostasis was secured and wound was closed over drain. Blood loss was significant and patient required two units of blood transfusion to keep blood haemoglobin in acceptable range. Patient was extubated on table. Postoperative period was uneventful.

Strap muscles of right side of neck and structures surrounding the mass found to be involved and dissected (Figure 4a and 4b).

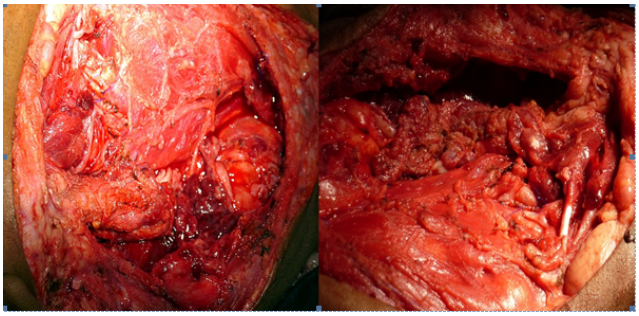

Figure 4a and 4b: The mass found to be involving surrounding structures and inoperable.

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Histopathological examination of excised tissue revealed (Figure 5A-5C) non circumscribed mass with typical areas composed of spindle shaped cells showing fascicular growth pattern with alternate hypo and hypercellular areas.

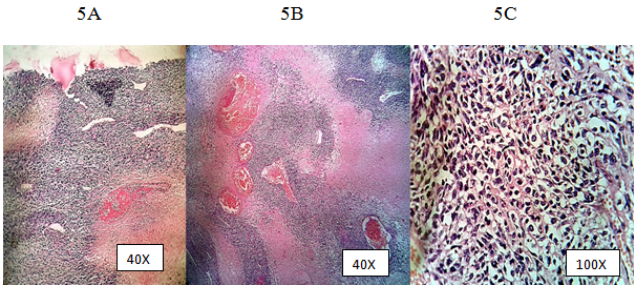

Figure 5: Histopathology of the tumour.

Individual cells had typical spindle or bucket handled shaped nuclei. There were areas of geographic necrosis and areas of haemorrhage. There were as such no areas of heterologous differentiation including skeletal muscle differentiation. Immunohistochemistry showed focal S-100 positivity and LCA negaitivity. MIB-1 labelling index was less than 5%. The diagnosis of malignant peripheral nerve sheath tumour was confirmed and the patient was referred to radiotherapy clinic. Sadly after a single course of radiotherapy patient suddenly developed high fever with chill, cough and profuse expectoration. Patient was admitted to intensive care unit. Pneumonia was diagnosed and the patient expired on the second day.

Discussion

Malignant peripheral nerve sheath tumor (MPNST) is a rare variety of soft tissue sarcoma of ectomesenchymal origin [1,2]. They arise from peripheral nerve branches5 or sheath of peripheral nerve fibers [9,10]. Head and neck are involved rarely (Table 1). Adults of 20-50 years of age are mostly involved [11].

Site of tumour	Percentage
Head and neck	04%
Chest wall and trunk	25%
Extremity Upper limb Lower limb	63% 29.2% 33.8%
Pelvis	08%

Table 1: Anatomical distribution of MPNST [3].

Most cases are large, more than 5 cm in diameter, fleshy and often necrotic [8]. MPNSTs are fusiform to globular in shape, not truly encapsulated and vary from white and firm to yellow and soft, depending on the absence or presence of necrosis [12]. The mass described in this study was firm, whitish and variegated in appearance (Figure 6).

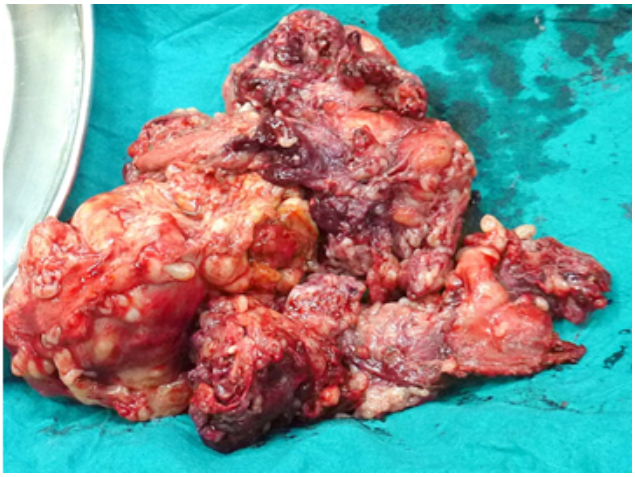

Figure 6: The debulked tumour.

Histologically MPNST may present with broad spectrum features without any specific diagnostic characteristic. Sections may reveal highly cellular spindle cell tumour with differentiation towards Schwann cells, nerve sheath and perineural cells with frequent mitoses and focal necrosis [13]. More commonly MPNSTs are formed of dense cellular fascicles alternating with myxoid regions [14]. The swirling arrangements of these areas are often referred as marbleized pattern. Nuclei may be spindle shaped, bucket shaped or fusiform. Features suggesting malignancy are invasion of surrounding tissues, invasion of vascular structures, nuclear pleomorphism, necrosis, and mitotic activity. MIB-1 labelling index may be used as marker of cellular proliferation and >5% cellular staining of MIB-1 proliferation marker has been considered as high grade tumor3. S-100 has been identified in approximately 50%-90% of MPNSTs, however the staining pattern has been noted to be both focal and limited to few cells [14]. In this patient, histopathological examination and immunohistochemistry pointed towards MPNST which did not corroborate with the FNAC finding. Leukocyte common antigen (LCA) staining was performed to exclude lymphoid malignancy as suggested by FNAC and immunoreactivity was absent. The fallacy of FNAC report was most probably due to sampling error as it was not image guided.

Clinical diagnosis of MPNST is difficult. A rapidly enlarging palpable fusiform mass in relation to a peripheral nerve in a patient with type 1 neurofibromatosis should bring suspicion to MPNST. Patients may present with dull aching or radicular pain, paresthesia or motor weakness. Magnetic resonance imaging (MRI) is the imaging modality of choice14. Large tumors (>5 cm), invasion of fat planes, heterogeneity, ill-defined margins, and edema surrounding the lesion are suggestive of MPNSTs14. For assessment of distant spread CT scan of chest, bone scan or FDG PET may be helpful. Fine needle aspiration cytology may not be adequate to establish initial diagnosis as tissue obtained may be inadequate.

The patient in this case presented with a neck swelling that was small and very slowly increasing in size for last 14 years and multiple small swellings in both extremity. There was a rapid increase in size of the swelling in previous 2 months. Clinically the extremity swellings were similar to neurofibromas. As those swellings were not symptomatic and not interfering with patient's normal activity the patient did not seek medical advice earlier. After admission, CT scan and MRI scan delineated the anatomical extension of the mass but failed to pinpoint the diagnosis. FNAC report pointed towards lymphovascular malignancy which was later found to be a sampling error. There is a probability of NF-1 positivity in this patient and maybe the presence of swelling for a long period (14 years) in a susceptible individual (NF-1 positive) induced malignant change in a benign swelling.

The treatment of MPNST is essentially surgical [3,7,15-20]. Radical resection and good three-dimensional clearance is essential for successful outcome. In case of head and neck tumours obtaining negative margins may be difficult because of relatively small space, proximity to vital structures and early haematogenous spread. Therefore these tumours have a poor prognosis and multimodality approach is more favorable [7,21-23]. Radiotherapy is also a good adjunctive treatment [3,7,17,19,24]. The oncology consensus group, as part of a uniform treatment policy for MPNSTs, recommends adjuvant radiotherapy for all "intermediate- to high-grade lesions and for low-grade tumours after a marginal excision" [17]. For extensive local spread the tumour in this case was found to be inoperable. For this reason the patient was referred to radiotherapy clinic postoperatively.

Several grading and staging systems have been described in literature for soft tissue sarcomas like MPNST and the French grading and National Cancer Institute grading systems are used more commonly [23]. The most commonly employed staging system is the American Joint Committee on Cancer Staging System for Soft Tissue Sarcomas [25,26]. Factors that correlate with prognosis [3, 7, 27] are sex, tumour location, depth and volume, tumour grade and cellular differentiation, presentation with either primary or recurrent disease or positive margin after surgery. Some studies indicated no association between tumour grade and prognosis [7,27]. Porter and co-workers [27] found NF-1 to be an independent indicator of poor prognosis in MPNSTs and recommended routine screening of these patients with FDG PET and or MRI for staging and controlling them at the earliest possible opportunity when the tumour volume is minimum.

Conclusion

MPNSTs are rare in head and neck region and carry an unfavorable prognosis. Diagnosis is difficult and fine needle cytology may be

misleading. Surgical resection with wide and negative margin is treatment of choice. Adjuvant radiation therapy should be delivered to improve local control and also may be beneficial for survival. Cases may present with large neck mass complicating airway management. Awake nasal fibreoptic bronchoscopy is safe in these cases.

Patient Consent

Taken

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