Case Report

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Oncology Section

Malignant Transformation of the Plexiform Neurofibroma of the Back: A Case Report

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ABSTRACT

Neurofibromatosis (NF) is characterised by multiple skin lesions distributed across the body and follows an autosomal dominant inheritance pattern. There are two main subtypes: NF type 1 (NF1) and NF type 2 (NF2), each with distinct clinical features. NF1 typically presents with numerous cutaneous neurofibromas, café-au-lait spots, plexiform neurofibromas, Lisch nodules, freckling in the axillary or inguinal regions, and optic gliomas. In contrast, NF2 is marked by bilateral vestibular schwannomas and central nervous system tumours such as meningiomas and ependymomas. A 69-year-old male presented with a swelling on his back. He had a malignant tumour that had transformed from plexiform NF to neurofibrosarcoma. A wide-excision biopsy of the swelling was done and a Malignant Peripheral Nerve Sheath Tumour (MPNST) was given as a diagnosis on Histopathological Examination (HPE). He underwent surgery for excision of the swelling. The patient had undergone Contrast-Enhanced Computed Tomography (CECT) of the thorax, abdomen and pelvis, showing cystic bronchial changes in the lower lobe and minimal in the posterior segment of the right lower lobe. Also, there was evidence of a large heterogeneous enhancing lobulated soft tissue density lesion in the subcutaneous plane of the neck and upper back. NF can be prevented from progressing if the malignant change is identified early. Surgical excision is the primary therapy; nevertheless, there is a greater chance of local recurrence, particularly in those suffering from NF1. The patient had undergone wide local excision with vacuum-assisted closure, which was followed by skin grafting.

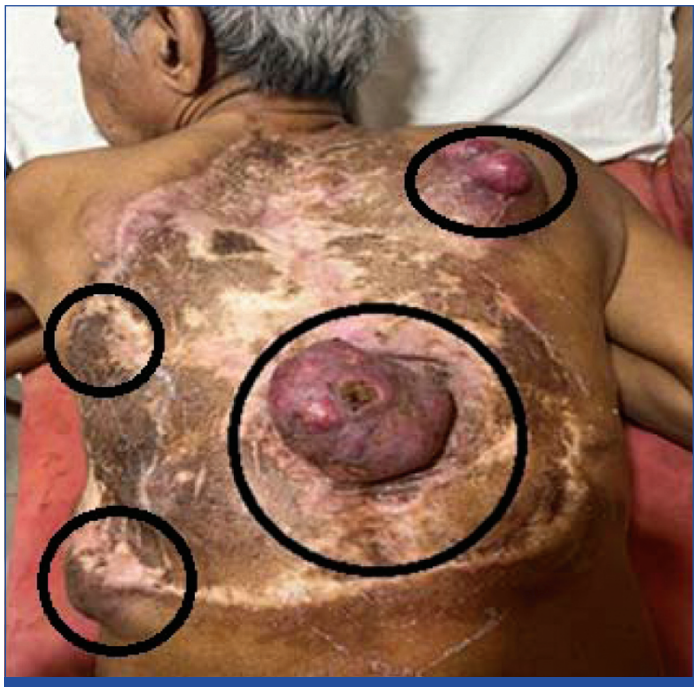
Keywords: Plexiform neurofibroma, Peripheral nerve sheath tumour, Sarcoma, Soft-tissue tumour, Swelling

CASE REPORT

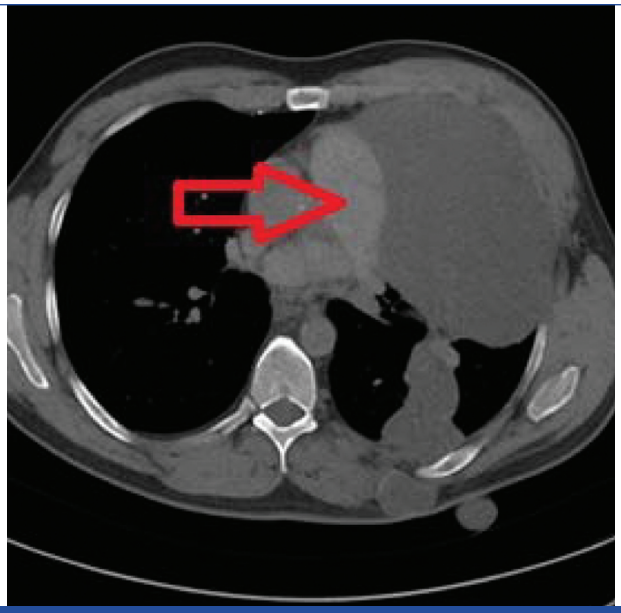
A 69-year-old male patient with underlying fibrosarcoma reported with multiple swellings all over the back for 10 months. The swellings over the back were painless but were bleeding with no history of fever and trauma. The patient was previously operated on four times in the past years and had taken 27 cycles of radiotherapy. After undergoing chemotherapy, he presented with a proliferating mass on the back. The mass measured 21×15×10 cm [Table/Fig-1]. In addition, the tumour was invading subcutaneous fat tissue. The patient was sent to a tertiary centre for further care because of an untreated infection. On examination by the surgical team, a large lump with necrotic skin was found on his back [Table/Fig-1]. Patient had undergone CECT thorax, abdomen and pelvis, showing cystic bronchial changes in the lower lobe and minimal in the posterior segment of the right lower lobe [Table/Fig-2]. There was evidence of a large heterogeneous enhancing lobulated soft tissue density lesion in the subcutaneous plane of the neck and upper back extending from C4-D7 level measuring approximately 18.6×5.9 cm [Table/Fig-3], which was suggestive of neoplastic aetiology. Another lesion with similar morphology in the mid back region involving the right thoracis spinalis muscle in the subcutaneous plane, measured 4.7×2.6 cm at the D10, D11 vertebral level. It caused erosion of the spinous process of the D10 vertebrae, which was most likely to be a metastatic deposit.

Whole spine screening revealed a similar lesion in the mid back region involving the right thoracis spinalis muscle measuring 4.7×2.6 cm at the levels of D10 and focal T2 hyperintense lesions noted in the L1, L2 and L3 vertebral body and a lesion at the L1 vertebral body extending into the pedicle, which was suspected as metastasis [Table/Fig-4].

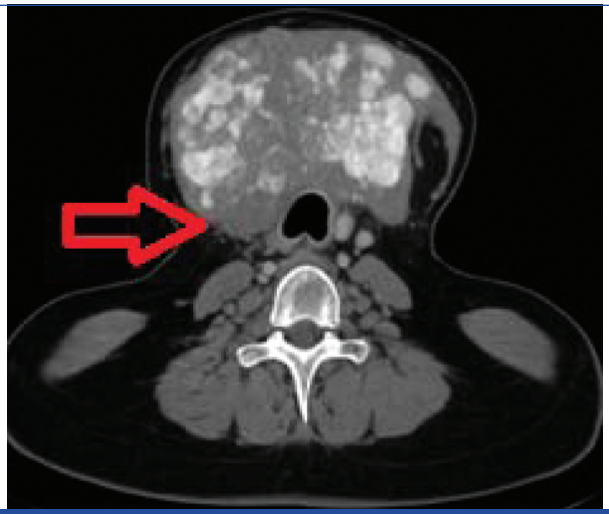
MRI spine dorsolumbar plain and contrast indicated that multiple T1-T2 intermediate density lesions were noted in the posterior back epicentred in the subcutaneous plane with both circumscribed margins showing mild diffusion restriction with evidence of homogeneous post-contrast enhancement and characteristics [Table/Fig-4]. Radiologically, the differential diagnosis of Plexiform Schwannoma (PS) was made, which is a rare histological subtype of schwannoma, characterised by a plexiform growth pattern and usually arising within the dermis and subcutaneous tissue. In terms of morphology, PS may present with conventional, cellular, or mixed features. However, its typical cellular architecture- characterised by hyperchromatic nuclei, increased mitotic activity, and a plexiform pattern- can resemble malignant features, particularly those associated with high-grade MPNSTs.



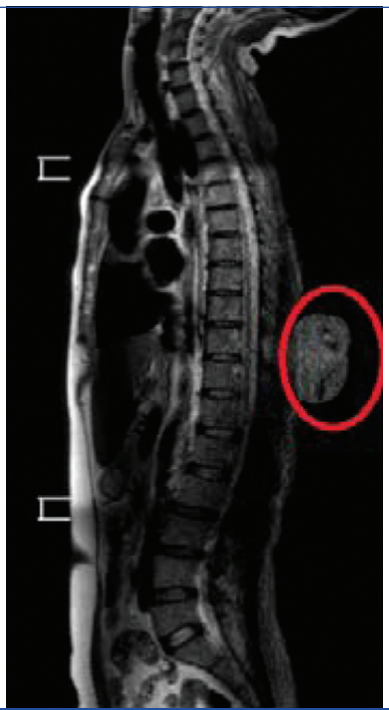
[Table/Fig-1]: Patient with multiple swellings on the back (fibrosarcoma).



[Table/Fig-2]: CECT showing cystic bronchial changes, which is evidence of a large heterogeneous enhancing lobulated soft-tissue density lesion.

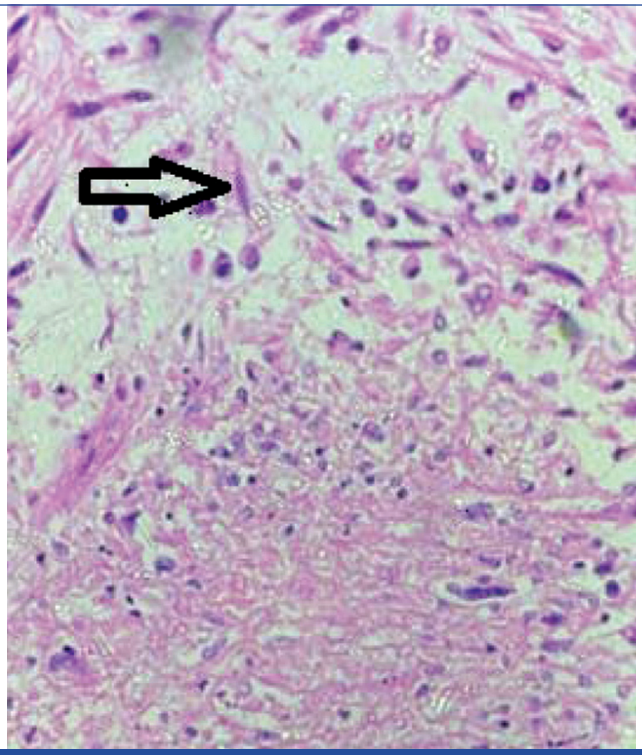


[Table/Fig-3]: Showing a large heterogeneous enhancing lobulated soft tissue density lesion in the subcutaneous plane of the neck and upper back extending from the C4-D7 level.

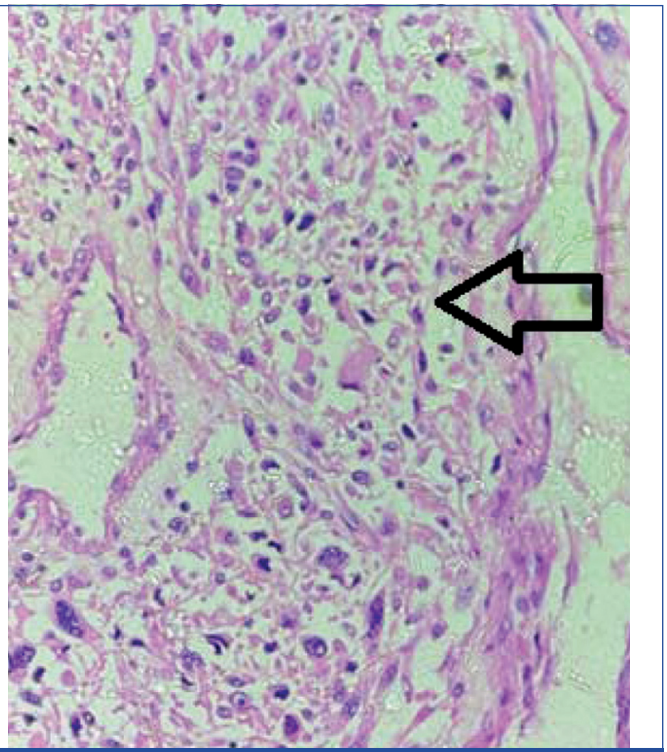


[Table/Fig-4]: Sagittal image of the Magnetic Resonance Imaging (MRI) spine shows an exophytic proliferative component at D10 vertebra.

A section from the tumour mass showed histopathological features suggestive of MPNST with Triton tumour-like differentiation [Table/Fig-5,6]. A portion of the tumour mass had histological characteristics that point to plexiform neurofibroma [Table/Fig-5,6]. The patient had undergone wide local excision with vacuum-assisted closure, which was followed by skin grafting.



[Table/Fig-5]: Uniform spindle cells with hyperchromatic, thin, wavy, or focally buckled nuclei (This is a haematoxylin and eosin stained slide in 20x magnification).



[Table/Fig-6]: Spindle cell formation with increased cellularity and scant cytoplasm disposed in sheets separated by thin fibrocollagenous stroma (This is a haematoxylin and eosin stained slide in 40x magnification).

DISCUSSION

Numerous cutaneous skin lesions that affect the entire body are a defining feature of NF. The inheritance pattern for this disease is autosomal dominant. NF type 1 (NF1) and NF type 2 (NF2) are the two subtypes of NF. Different traits can be used to distinguish between the two categories. While patients with NF2 are characterised by

bilateral vestibular schwannomas and Central Nervous System (CNS) tumours, including meningiomas and ependymomas, individuals with NF1 may present with numerous cutaneous neurofibromas, cafe-au-lait spots, plexiform tumours, Lisch nodules, axillary or inguinal freckling, and optic gliomas. The major therapies for NF include ongoing monitoring and, where necessary, medical intervention. There is no particular therapy for NF. Neurofibroma patients experience widespread neurocutaneous lesions across their bodies. In individuals with NF, delayed diagnosis and follow-up may lead to malignant transformations of the NF. Neurofibrosarcomas, often referred to as malignant schwannomas, neurogenic sarcomas, or MPNSTs, can develop from plexiform neurofibromas [1].

PS is a rare histological subtype of schwannoma, characterised by a plexiform growth pattern and most often found in the dermis and subcutaneous tissues. Its morphology can vary, presenting as conventional, cellular, or a combination of both. However, the commonly observed cellular form- marked by hyperchromatic nuclei, elevated mitotic figures, and plexiform architecture- may mimic malignancy, particularly resembling high-grade MPNSTs [2].

The neurocutaneous lesion became bigger and larger as it underwent a malignant change. Tumour cells will invade the surrounding mass as the lesion grows, potentially jeopardising and harming its blood supply. Ischaemic alterations cause the skin and surrounding tissue to eventually die [3]. As a result, the tumour forms a fungating mass and begins to ulcerate. According to experts, there is a 4.6% lifetime chance of malignant transformation [4]. Research indicates that individuals with cutaneous neurofibroma are three times more likely to acquire an MPNST [4,5].

An uncommon genetic condition that is inherited in an autosomal dominant manner is NF. Usually, it results in the development of other body areas and benign nerve tumours. NF1, NF2 and Schwannomatosis (SWN) are the three varieties. Many cutaneous neurofibromas, cafe-au-lait spots, plexiform tumours, Lisch nodules, axillary or inguinal freckling, and optic gliomas are characteristics of NF1. The illness might appear anywhere in the system. It has the potential to develop into several malignancies, including the far more prevalent MPNSTs, brain tumours, and breast cancer [6].

It is challenging to differentiate between PN and MPNST in clinical presentation because of their similar appearances. Some PN patients live their whole lives without experiencing any symptoms. An earlier stable neurofibroma with a history of fast development raises the possibility of malignant change. There is a significant chance that any lump larger than 5 cm is malignant. If we come across masses larger than 5 cm, a proper assessment needs to be conducted. Eliminating malignant tumours is the most suitable and crucial course of action. A delayed diagnosis might result in distant metastases and invasion of nearby tissues. Additionally, malignant evolution of soft-tissue tumours is indicated by ulcerated, large, and surrounding erythematous appearances [7].

MPNST exhibits extremely aggressive behaviour. Surgical resection is the primary treatment for MPNST, and it is uncertain how adjuvant treatments would affect prognosis [8,9]. In order to achieve negative margins, a thorough surgical resection is the only viable therapy. Controlling local recurrence and, eventually, patient survival depend on having an adequate margin. Compared to patients with positive margins, complete resection has a greater five-year survival rate and a lower recurrence rate [10,11]. According to reports, NF1 status is a risk factor for MPNST prognosis [11,12].

Iqbal F et al., operated on a 38-year-old female patient diagnosed with NF-1; her left side had a single, asymmetrical, cystic growth that was becoming bigger. The patient had a 6 cm tumour surgically removed; upon histological analysis, the cancer was determined to be MPNST. Due to its rarity, this tumour was very difficult to diagnose and treat [13]. A 47-year-old man having type 1 NF with haemorrhage because of a stomach subepithelial lesion was described by Hwang HS et al. After confirming the histological appearance and immunohistochemistry profiles, it was determined that the operation he had undergone was for MPNST, rather than a gastrointestinal stromal tumour. The big tumour also infiltrated the diaphragm and spleen. Additional chemotherapy was given in conjunction with radical surgery [14].

CONCLUSION(S)

NF1 is mostly linked to MPNST. The majority of PNs develop into cancerous lesions. Malignant alterations and other issues can be avoided with early management. Treatment choices are tailored individually, taking into account factors such as the patient's age, tumour location and size, presenting symptoms, and general health status. In individuals with NF1, appropriate monitoring may increase survival and limit malignant PN transformation. Radiotherapy and chemotherapy limit the effectiveness of neurofibroma. Surgical excision is the primary therapy. Surgery aims to remove as much of the tumour as possible without causing further nerve damage.

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