

# Malignant Peripheral Nerve Sheath Tumour In Neurofibromatosis Type 1

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## Abstract

Malignant peripheral nerve sheath tumours (MPNST) are rare. We present a case of a 30-year-old African male patient with neurofibromatosis type I with a MPNST involving the sciatic nerve. A subtotal resection of the tumour was done with preservation of the sciatic nerve with good neurological outcome. Complete tumour resection is advocated. However, due to sacrifice of major nerves to achieve a tumour-free margin and the reluctance of patients to undergo amputation, subtotal resection and adjuvant radiotherapy provides the best alternative in treating this aggressive disease.

## INTRODUCTION

Malignant peripheral nerve sheath tumours (MPNST) are rare with an incidence of 0.001% of the general population<sup>1</sup>. There is a clear association between MPNST and Neurofibromatosis Type 1 (NF 1), the estimated risk being up to 4600 times greater than in the general population<sup>2</sup>. We present a rare case of a MPNST involving the sciatic nerve in a young African male with NF 1 to highlight the surgical approach and controversies in the management of this soft tissue sarcoma.

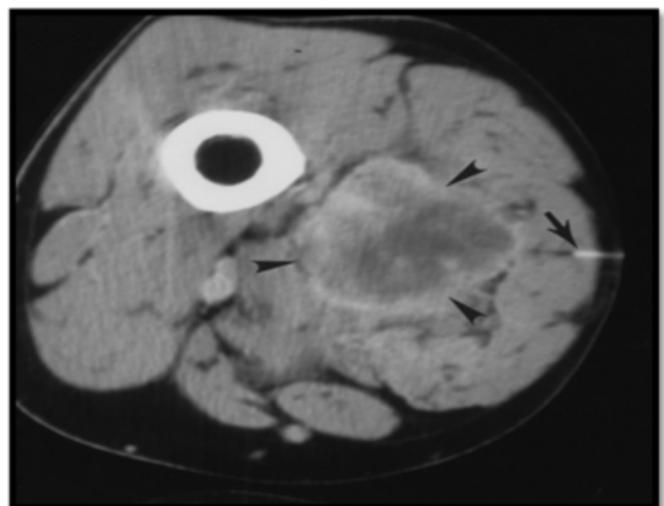
## CASE STUDY

A 30-year-old African male presented with a slow growing mass in the left posterior thigh of 3-year duration associated with pain and paraesthesia. Clinical examination revealed multiple cutaneous neurofibromas and a posterior thigh mass approximately 10cm by 12cm in size, firm, non-tender, non-pulsatile and fixed. There were no associated vascular or neurological deficits and the patient walked with an antalgic gait. Plain radiographs demonstrated soft-tissue swelling with no bone involvement. Ultrasound examination revealed a large mass with multiple hypoechoic areas with no increase in vascularity. Contrast enhanced CT scan (figure 1) showed a well encapsulated solid mass with cystic areas involving the posterior compartment of the left thigh arising from the sciatic nerve with no evidence of vascular or bone involvement. The patient underwent a surgical exploration at which time the tumour was found to be well encapsulated and adherent to the sciatic nerve (figure 2). A subtotal resection of the tumour was performed with preservation of

the nerve fascicles adherent to the tumour capsule. The sciatic nerve was preserved. Histological evaluation revealed a high-grade MPNST with areas of necrosis with S100 positivity on immunohistochemical staining. In view of the positive margins and high tumour grade, the patient was planned for adjuvant radiotherapy. Post-operatively, there was no neurological deficit and his symptoms had resolved.

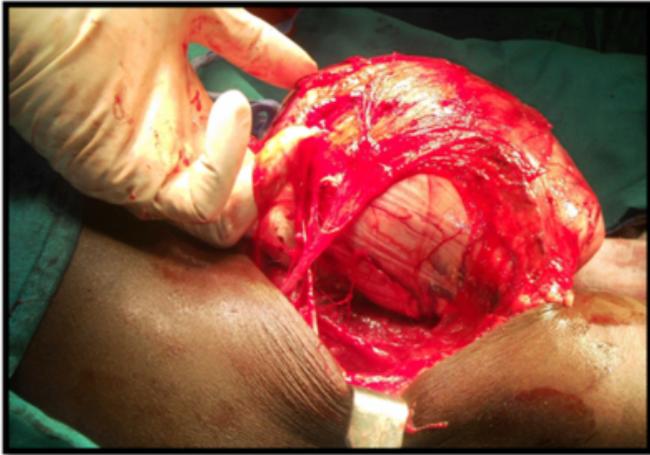
## Figure 1

Figure 1. CT scan demonstrating a well-encapsulated solid mass with cystic areas involving the posterior compartment of the left thigh arising from the sciatic nerve



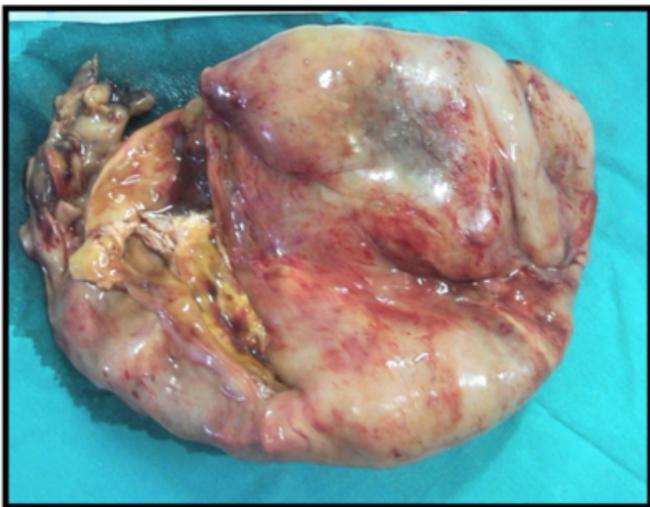
**Figure 2**

Image 2: Intra-operative view: Well encapsulated tumour adherent to sciatic nerve



**Figure 3**

Image 3: Specimen: 15cm by 12cm well-encapsulated tumour with areas of necrosis and cyst formation



## DISCUSSION

MPNST is a term that was coined by the WHO to encompass all malignant neurogenic tumours involving the major and minor peripheral nerves<sup>2</sup>. The pathological criteria for malignancy include invasion of surrounding tissues by tumor cells, vascular invasion, marked nuclear pleomorphism, necrosis, and the presence of mitoses. The minimum histological examination should comprise sections stained with conventional stains including H&E and reticulin. In addition, immunohistochemical stains for S100 protein, the skeletal muscle markers desmin and myogenin, and a proliferation marker (MIB1) are required<sup>3</sup>.

The diagnosis of a MPNST rests on a high index of clinical

suspicion. The presence of a solid mass with fixity and neurological deficit may suggest a malignant tumour. MRI is the imaging modality of choice displaying exquisite soft-tissue detail and defining the relationship of the tumour to neurovascular structures. Features suggestive of malignancy include areas of haemorrhage or necrosis, heterogeneous enhancement, and cystic areas; however, MRI does not reliably distinguish malignant from nonmalignant tissue within a neurofibroma<sup>4</sup>. PET scanning is currently reserved for staging and detection of recurrent disease<sup>5</sup>.

Surgery remains the mainstay of therapy for MPNST. The goal of surgery is limb-preservation with complete tumour resection with negative margins. If the tumour is deemed irresectable due to involvement of a major nerve, then subtotal resection with adjuvant radiotherapy is an alternative option to primary amputation. The recurrence rate following surgery is 32% to 65% and routine adjuvant radiotherapy is recommended in all MPNSTs irrespective of margin clearance or tumor grade<sup>2</sup>. Radiotherapy is preferred in the postoperative period due to the lower incidence of wound infection and less distortion of tissue planes facilitating dissection<sup>6</sup>. Amputation may be indicated for extensive tumours and for MPNSTs which recur after apparently adequate excision<sup>3</sup>. Chemotherapy is generally confined to the treatment of metastatic disease. The overall survival benefit with chemotherapy is marginal<sup>7</sup>. The reported overall 5-year survival rate for MPNST with NF I is 10%. Poor prognostic factors for MPNST include tumor size, histological grade, positive resection margins and NF I.

MPNST carries the highest recurrence rate for soft-tissue sarcoma surgery; thus, the best chance of cure is offered at the initial surgery where complete tumour resection is advocated. However, due to sacrifice of major nerves to achieve a tumour free margin and the reluctance of patients to undergo amputation, subtotal resection and adjuvant radiotherapy provides the best alternative in treating this aggressive disease<sup>8</sup>.

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