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CASE REPORT
Peripheral Nerve

# Diffuse Type Neurofibroma of the Forearm

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**Summary:** Nerve sheath tumors comprise 5% of soft tissue masses of the upper limb in adults. Neurofibromas are divided into three types: localized, diffuse, and plexi- form. The diffuse type is rare and is typically found in the head and neck region. We present a rare case of diffuse type neurofibroma found in the forearm, presented to our clinic as a slowly enlarging mass of the left forearm of 3 years duration. The lesion was suspicious in the magnetic resonance imaging, and biopsy revealed diffuse type neurofibroma. We opted for total excision of the lesion that was found to be not possible due to involvement of the major nerves. The final pathology report showed no malignancy. Nerve tumors of the upper limb can be either benign or malignant. Neurofibroma associated with neurofibromatosis has malignant potential. The diffuse type is rare, and it most commonly occurs in the head and neck region. It has a low malignant transformation rate. Magnetic resonance imaging is the diagnostic modality of choice; however, it can be inconclusive. Biopsy should be taken to confirm the diagnosis and plan for management. Our case was managed by near total excision in order to preserve the major forearm nerves because of high clinical suspicion.
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Soft tissue masses of the forearm can arise from the comprising structures (fat, muscles, nerves, or connective tissue). Nerve tumors constitute around 5% of upper extremity tumors in adults.[1,2] Nerve tumors are divided into schwannomas and neurofibromas. Neurofibroma can occur sporadically or as a part of neurofibromatosis (NF).[3] It is classified into three types: localized, diffuse, and plexiform.[4] The diffuse type is rare, and it typically involves the skin and subcutaneous tissues of the head and neck.[5] In this article, we describe a rare case of diffuse type neurofibroma aggressively involving the tissues of the forearm in a 25-year-old patient. There were no other lesions or swellings other than café au lait spots distributed all over his trunk, suggesting neurofibromatosis type I.

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## CASE PRESENTATION

A 25-year-old Nepalese man, who is a laborer and is right-handed, presented to our clinic complaining of a huge swelling in his left forearm that had been growing slowly for the last 3 years with no associated pain, weakness, or paresthesia. The patient did not have any comorbidities, and his surgical history was only remarkable for a left distal ulnar fracture which he sustained 10 years before presentation. On physical examination, the swelling extended from the left elbow down to the distal third of the forearm. It had two distinguishable components: a solid one that was attached to the deep structures, and a soft one that was blended with the subcutaneous tissue. There was no change in the overlying skin, and no motor or sensory deficits. MRI showed non-homogeneous intermediate signal intensity on T1. [**See figure, Supplemental Digital Content 1,** which displays MRI showing the described lesion (yellow arrow). **http://links.lww.com/PRSGO/C44**.] Postcontrast images showed significant enhancement. The differential diagnoses suggested by radiology were vascular malformation, chronic TB infection, and unusual soft tissue sarcoma. Two incisional biopsies were taken from the solid and the soft variants of the tumor. The pathology report showed diffuse type neurofibroma. Despite proper hemostasis, the procedure was complicated by a large hematoma, which

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mandated surgical evacuation and hemostasis. Because the clinical findings were suggesting malignant behavior, excisional biopsy was planned to ensure proper diagnosis. Intraoperatively and under ×4 magnifying loops, the mass was seen violating the fascia and muscles (Fig. 1). The solid element of the mass was adherent to the bone, causing a unicortical erosion. We were able to debulk most of the tumor mass but not completely because it was incasing both the median and ulnar nerves. Postoperatively, the patient had full hand function. The pathology report showed diffuse type neurofibroma with no malignancy or atypical features. The patient was discharged and referred to the genetics clinic for neurofibromatosis counseling, and he is under regular follow-up.

## DISCUSSION

Nerve tumors of the upper extremity include benign peripheral nerve sheath tumors (BPNSTs) and malignant peripheral nerve sheath tumors (MPNSTs).[6] BPNST can be broadly divided into neurofibromas and schwannomas. Neurofibromas are complex tumors composed of axonal processes, Schwann cells, fibroblasts, perineurial cells, and mast cells. In sporadic and syndromic cases, a deletion in the NF-1 gene in the Schwann cell lineage is sufficient to generate tumors.[7] Neurofibroma is the most common tumor of the peripheral nerve sheath. It affects men and women equally, with no racial or ethnic predilection. Age of onset is highly variable, and localized lesions most commonly occur in adults aged 20–40 years. The diffuse and plexiform types occur more frequently in children.[8]

Malignant transformation of neurofibromas is rare. The risk is increased if there is associated NF, especially with the plexiform type.[9,10] MPNSTs may occur in 2%–13% of patients with NF type I, compared with 0.001% of the general population.[11,12] Symptoms of malignant transformation are rapid growth, pain at night, and tumor size more than 5 cm.

The diffuse type of neurofibroma is a distinct and rare type of neurofibroma. It is commonly found in the head and neck region. In around 60% of cases, it is associated with NF type I. It typically presents in young adults as an ill-defined plaque with subcutaneous thickening. Malignant transformation is extremely rare. On gross examination, diffuse type neurofibromas appear as ill-defined dermal/subcutaneous thickening extending into subcutaneous tissue and fascia.[10]

Evaluation of a solitary soft tissue mass in the upper extremity should begin with plain radiographs, then computed tomography or magnetic resonance imaging, with magnetic resonance imaging being the most useful imaging modality.[13] None of the imaging modalities can accurately distinguish nerve sheath tumors from others. Malignancy can be predicted through the invasion pattern that appears in the imaging study.[14] F-18 fluorodeoxyglucose positron emission tomography has been recently studied for the detection of MPNSTs.[11] Failure to identify the lesion through imaging would necessitate the use of core biopsy for diagnosis.

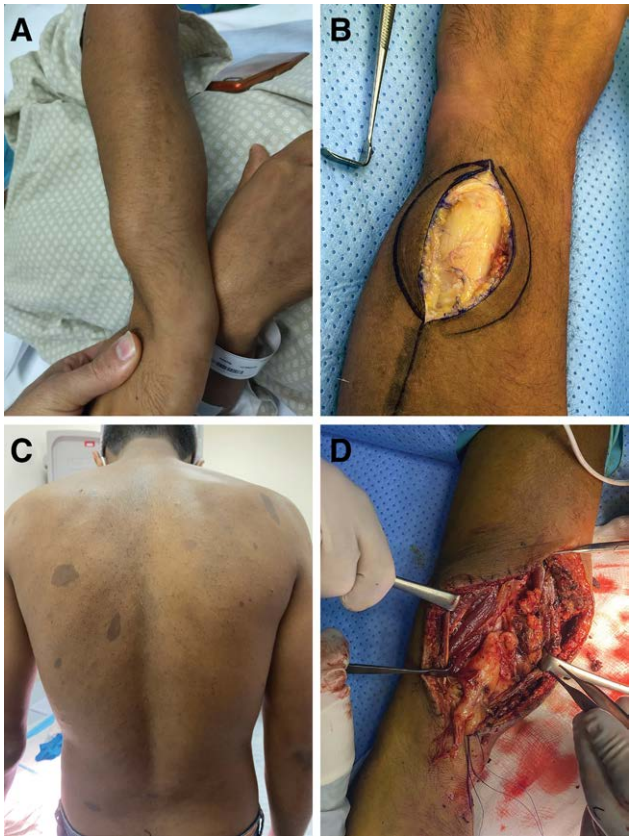


**Fig. 1.** A, Clinical appearance of the tumor on the left forearm with soft and hard components. B, Café au lait spots on the trunk. C, Intraoperative image of the tumor at the time of incisional biopsy. D, Intraoperative image of the tumor at the time of near total excision.

The first step in managing a soft tissue mass in the upper limb is through incisional biopsy to determine the origin of the tumor, then to proceed according to the final pathology report. BPNSTs can be observed if they remain stable in size and are asymptomatic, but for most cases, complete surgical excision is the preferred treatment. Local recurrence is extremely rare. There are currently no alternative therapies for cutaneous neurofibromas. Surgical management of BPNSTs is with marginal excision.[1] Neurofibromas classically intertwine with the underlying nerve, making intraneural dissection and fascicular preservation not always possible.[9,15] These patients require monitoring to detect any malignant transformation or recurrence. If the involved nerve is of functional importance, biopsy would be enough to exclude malignancy. Otherwise, the nerve can be cut with the involved segment and repaired primarily or through a nerve graft. Cutaneous nerves can be compromised.[1] MPNSTs require wide excision, and the management should be approached by a multidisciplinary cancer team. Amputation should be considered for large or recurrent tumors. Radiotherapy for MPNSTs may provide local control but has not been shown to prolong survival.[11,16]

Our patient had diffuse-type neurofibroma in the forearm with no other tumors elsewhere in the body, and

it was associated with café au lait spots, suggesting neurofibromatosis type I. Diffuse-type neurofibroma usually appears in the head and neck region and in the trunk. It has not been reported in the literature to be found in the forearm. The tumor had soft and solid components, which is not a characteristic of diffuse type neurofibroma. It was violating the deep facia, muscles, and bones. It encircled the ulnar and median nerves. The high clinical suspension of a malignant component made us opt for tumor debulking even after having a biopsy report as benign neurofibroma. To preserve the main forearm nerves, we were not able to excise the entire tumor, but most of it was removed. This type of tumor is highly vascularized, and our case was complicated by hematoma formation twice after the initial biopsy, which was managed surgically the first time, and conservatively the second.

Our main purpose of writing this article is to draw attention to how to manage such a tumor according to the literature provided, as it is rarely encountered. The patient will be followed up in our clinic to follow his disease course and to check for any recurrences.

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