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# Synovial Sarcomatous Transformation in Recklinghaussen Disease: A Case Report

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**Abstract** **Case Report**

Malignant sarcomatous transformation of neurofibromas MSTN are a major cause of mortality in patients with NF1, its optimal management and final prognosis depend on early and accurate detection. Through a case report and a literature review, we will discuss the specificity of sarcomatous transformation in Neurofibromatosis type 1.

**Keywords:** MSTN, Neurofibromatosis, chromosome 17.



## INTRODUCTION

Neurofibromatosis (NF), is an autosomal dominant disorder that is caused by a defect or mutation of a tumor suppressor gene located on chromosome 17 [1]. The most common type of NF is NF type 1 (NF-1).

Although malignancies associated with NF-1 are rare, the condition is associated with an increased risk of malignant peripheral nerve sheath tumors (MPNSTs), optic gliomas, and leukemia.

A synovial sarcoma is a spindle cell sarcoma of soft tissue of uncertain histogenesis. It is a rare tumor, accounting for 2% of all sarcomas [3]. They appear as deep tissue masses in the extremities or the trunk.

Herein, we report a case of an MPNST of the skin type synovial sarcoma in a patient with NF-1 seen at Ar-razi Hospital in Marrakech. The diagnosis was suspected through imaging and confirmed by anatomopathological study.

## CLINICAL HISTORY

A 28 years old female patient, with a history of neurofibromatosis type I since childhood presented with low-grade fever and unintentional loss of weight of 7kg over 5 months. She also complained of increasing size of a right brachiate neurofibroma.

Clinical examination revealed multiple cutaneous neurofibromas and café-au-lait spots on her face (Figure 1), body and limbs. Clinically palpable soft tissue masses, were felt on her thoracic wall and right arm with no obvious skin changes.

The ultrasound had revealed a subcutaneous mass, quite well-defined, with a heterogeneous structure on ultrasound, showing color Doppler signal.

CT had revealed a subcutaneous well defined mass in the right arm, spontaneously isodense presenting an heterogeneous enhancing after contrast (Figure 2).

Magnetic Resonance Imaging (MRI) of the right arm (Figure 3) demonstrated a heterogeneous soft tissue mass measuring 11 × 11 × 32 cm, seen in the anterior compartment, with tumoral heterogeneous signal and peripheral rim enhancement. Peri-lesional oedema was seen in the adjacent soft tissues. Findings raised suspicion for malignant transformation of a preexisting neurofibromatosis. Subcutaneous and cutaneous soft tissue nodules demonstrating homogeneous T2 signal and enhancement were noted, in keeping with benign neurofibromas.

The patient underwent an ultrasound guided biopsy of the lesion. Histopathological findings from the tumor showed interlacing bundles of spindle cells compatible with a synovial sarcoma

After diagnosis of the mass as MPNST, complete surgical excision was planned.

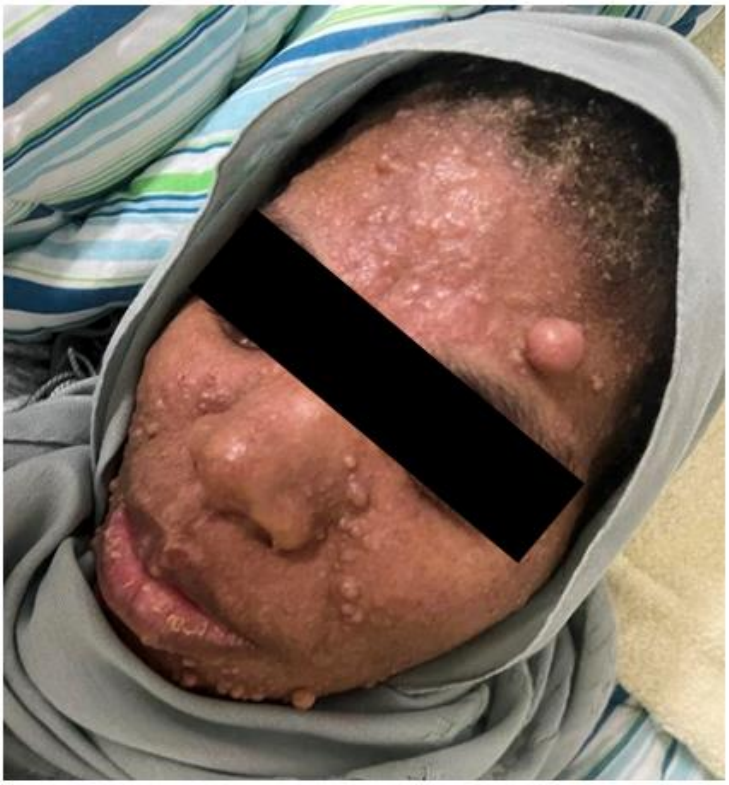

**Figure 1: Clinical imaging of the patient**

Clinical imaging showing multiples facial neurofibromas.

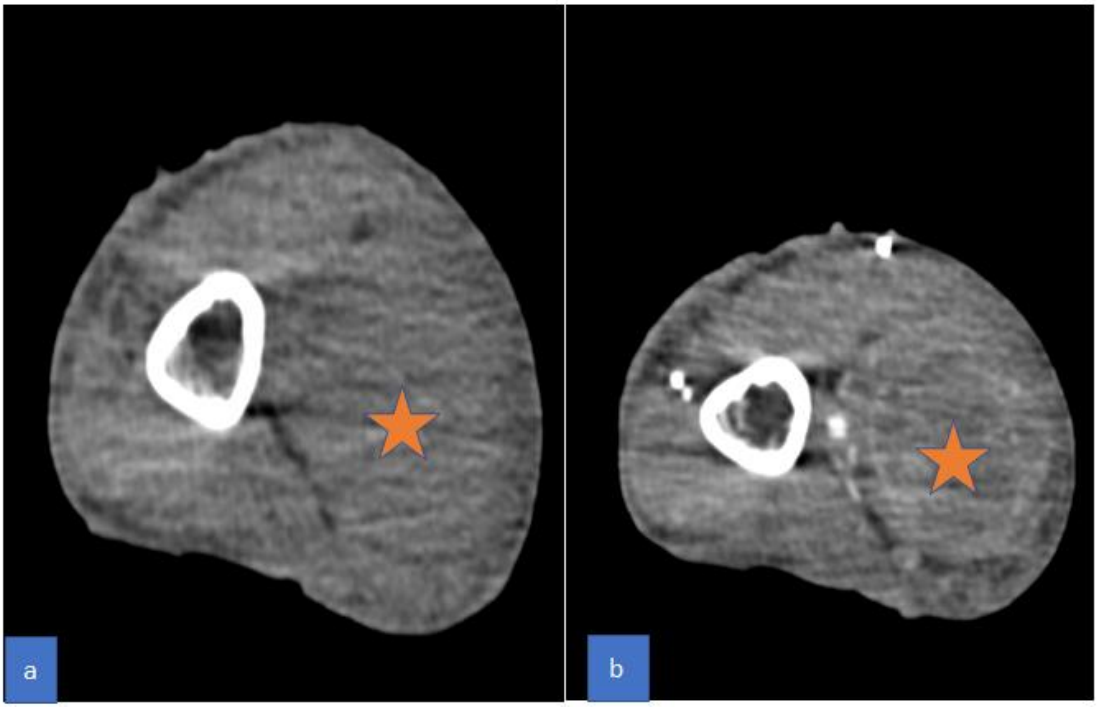


**Figure 2: A Non contrast CT, b. Constrast CT of the right arm**

Well defined isodense subcutaneous masse, spontaneously isodense with heterogenous enhancement defining necrotizing zones.

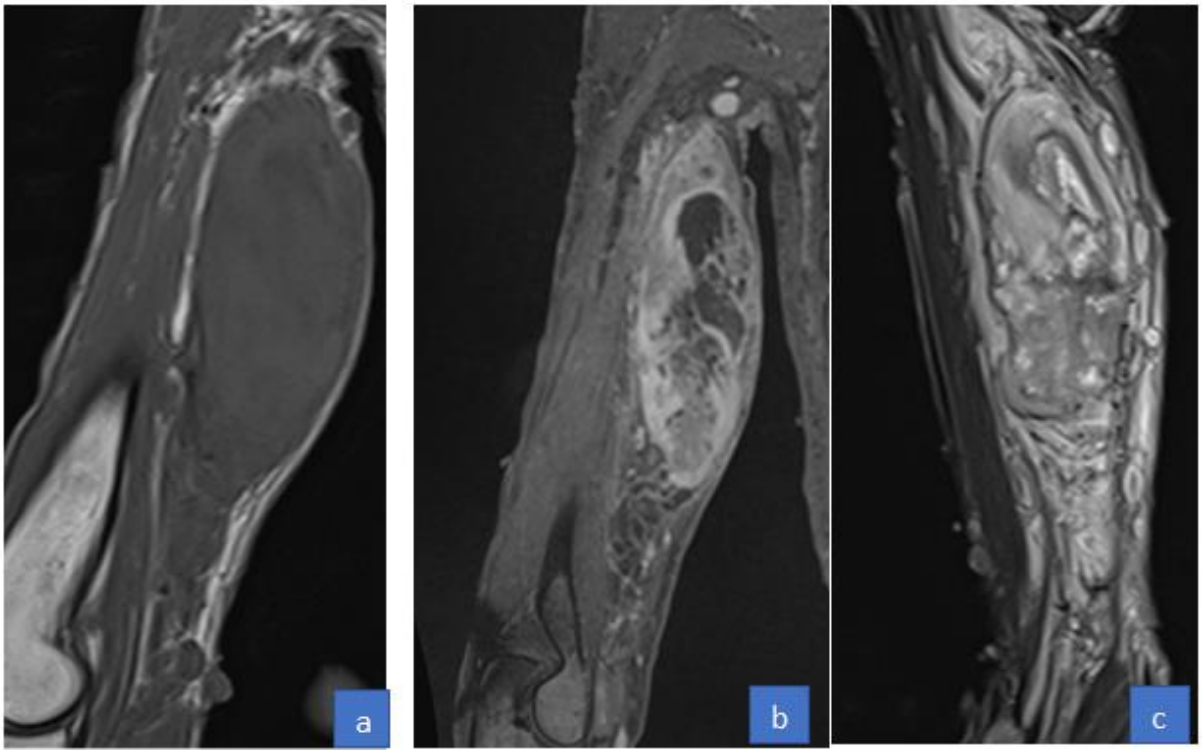


**Figure 3: MRI of right arm a. T1, b T1 with contrast, c. STIR**

An heterogeneous soft tissue mass measuring 11 × 11 × 32 cm, seen in the anterior compartment, with tumoral heterogeneous signal, peripheral rim enhancement and peri-lesional oedema.

## DISCUSSION

Neurofibromatosis type I (NF1) is a common autosomal dominant multisystem disorder, and its classical features are "café au lait" spots, iris Lisch nodules, neurofibromas, and skin fold freckling. Neurofibromas are the most common tumor associated with NF1, and among the three subtypes (cutaneous, subcutaneous, and plexiform) plexiform neurofibroma (PN) represents a major cause of morbidity and mortality [1].

Malignant sarcomatous transformation of neurofibromas MSTN are a major cause of mortality in patients with NF1, and optimal management and final prognosis depend on early and accurate detection of malignant transformation. Clinical indicators of malignant degeneration include persistent or increasing pain, increasing tumor size, and neurologic deficits, but these findings may be seen in both benign and malignant lesions, and surgical biopsy is often necessary to exclude malignancy [4].

Radiographs are of limited value except in the evaluation of potential aggressive bony features. Ultrasonography (US) is easily accessible and cost effective, however it is also of limited value in evaluating deeply located lesions. Computed Tomography (CT) is effective in delineating the extent of the tumor, identify bony involvement and good for preoperative planning.

MRI remains the best imaging modality for evaluating MPNSTs due to its high soft tissue resolution and ability to further characterize different components of the tumor and delineate margins for surgical planning. Imaging features suggestive of malignant transformation include an enlarging mass, presence of peripheral enhancement, perilesional oedema, and increased tumor heterogeneity [5].

- **Increased largest dimension of mass**: Interval increase in the largest dimension of a known neuroofibrma, especially if associated with high-risk clinical manifestations such as pain at rest, increasing or acute onset of pain, neurologic symptoms and radiculopathy, should raise suspicion for malignant transformation. The interval increase in size is probably due to rapid growth in malignant tumor. The average size of a MPNST is usually above 5cm.
- **Increased tumour heterogeneity:** Malignant transformation of a tumor can result in hemorrhage or necrosis, which is reflected as heterogeneity on T1-weighted images, and intra-tumoral cystic change on T2-weighted images. Some studies reported that a '**target sign'** may be helpful in differentiating between benign and malignant neurofibromas. A target sign is described as a central hypointense region seen on T2-weighted images, surrounded by a rim of T2 hyperintensity. This sign has been

commonly associated with localized benign neurofibroma, corresponding to a central area of hypercellular stroma surrounded by myxomatous tissue on histopathology [6]. Loss of the target sign may be suggestive of malignant transformation.

- **Presence of heterogenous enhancement:** Development of heterogenous enhancement due to necrosis and hemorrhage, is a sign that is not often observed in benign neurogenic tumors and should raise suspicion for malignant transformation.
- **Peri-lesional oedema:** Benign neurofibroma tend to be more well-defined and are usually surrounded by a capsule. Peri-lesional oedema occurs when malignant transformation of an existing peripheral nerve sheath tumour occurs with marginal infiltration of tumour cells into surrounding soft tissue, causing a localized reaction [3].

Complete surgical excision is a treatment option in cases of MPNST [1]. Response to chemotherapy and radiotherapy is known to be poor, and chemotherapy can be used only for metastatic diseases. Our patient underwent surgery, after which no adjuvant therapy was administered. The most important prognostic factor is tumor size, which is typically >5 cm at presentation. Other factors include tumor grade, surgical margin, local recurrence, histological malignancy, cell polymorphism, and mitotic activity [7].

## CONCLUSION

Differentiating malignant transformation of benign tumours continues to be a challenge in clinical practice with radiology playing an important role in answering clinical questions; MRI continues to be the modality of choice for characterizing and delineating the target lesion.

Imaging features of malignant transformation guide radiologists in sieving sinister lesions from benign entities; high risk factors would include: increased largest dimension of mass, increased tumour heterogeneity, peripheral enhancement and perilesional oedema, as in our case report.

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