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## CASE REPORT

# MRI evaluation of diffuse subcutaneous neurofibroma of the lower limb in a low resource setting

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### ABSTRACT

An unusual type of neurofibroma predominantly seen in children and young adults is diffuse neurofibroma. We present a 25-year-old female with recurring soft tissue masses in her right lower limb. MRI showed areas of  $T_1$  iso-intensity and  $T_2$  hyperintensity relative to skeletal muscle within the subcutaneous fat. These masses show marked enhancement post gadolinium administration. Histological examination of the excised mass showed diffuse neurofibroma. The rare nature of this tumour and the limited literature describing the imaging features make the diagnosis relatively difficult for a radiologist.

### CLINICAL PRESENTATION

A 25-year-old African female presented at the MRI suite with several years history of a progressively enlarging right calf and ankle swelling. She was referred by the orthopaedic surgeon for a musculoskeletal MRI of the right calf with a clinical diagnosis of a soft tissue sarcoma. On examination, an ill-defined mass was noticed at the posterior right calf. It was warm, firm and not painful or tender to touch. A similar mass was observed at the right ankle. There was an excision of a similar swelling 10 years prior to representation.

Differential diagnoses included diffuse subcutaneous neurofibroma and haemangioma.

### INVESTIGATION/IMAGING

Routine haematology and blood chemical pathology tests were normal. She has no significant past medical history or family history of neurocutaneous disease.

Plain radiograph (Figure 1) showed two oval well-defined homogenous soft tissue masses in the posterior right calf and at the ankle. No abnormal calcification or phleboliths were seen. The overlying skin and adjacent bones were normal.

Serial axial and coronal  $T_1W$  ( $T_1$  weighted),  $T_2W$  ( $T_2$  weighted), STIR, PD and sagittal  $T_2W$  as well as post-contrast axial MRI images were acquired. These show a well-defined oval-shaped mass which is iso-intense on  $T_1W$  and

hyperintense on  $T_2W$  sequences in the subcutaneous tissue of the right calf, displacing the adjacent muscles anteriorly. This mass measures 8.3 cm by 4.0 cm by 5.3 cm in longitudinal, anteroposterior and transverse dimensions, respectively. (Figure 2) A similar but smaller mass was seen in the subcutaneous tissue in the lateral aspect of the right foot. These masses show marked and bright heterogeneous enhancement after intravenous administration of paramagnetic contrast agent (Gadolinium DTPA).

Surgical resection of the mass was performed and subsequent histological examination reported a benign nerve sheath neoplasm composed of spindle cells disposed in sheets on a variably fibromyxoid stroma. The spindle cells have wavy to curved elongated nuclei with scant eosinophilic cytoplasm. (Figure 3).

Figure 1. Plain radiographs showing homogenous soft tissue masses over the right ankle (a) and calf (b).

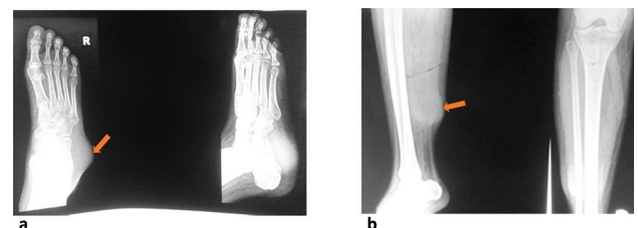
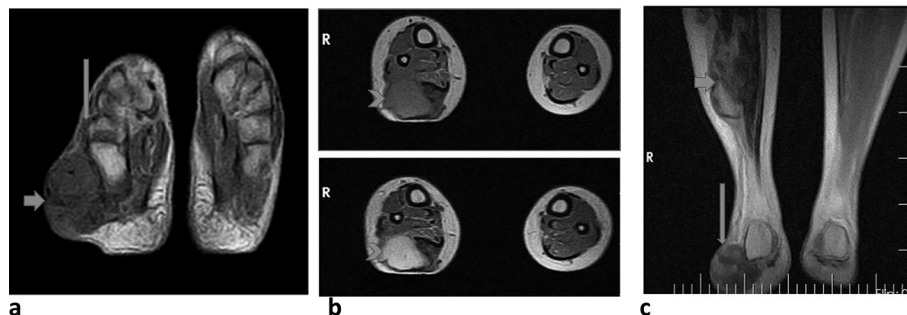


Figure 2. (a) Pre-contrast spin echo axial  $T_1$  weighted MR image (TR/TE, 904/16) shows a bulging mass (short arrow) in the subcutaneous tissue over the right ankle which is iso-intense to adjacent muscle (long arrow). (b) Pre- and post- contrast spin echo axial  $T_1$  weighted MR images (TR/TE, 904/16) show enhancement of the right posterolateral calf mass (arrow heads). (c) Coronal  $T_2$  weighted fat-saturated MR image (TR/TE, 2000/32) shows a bulging mass (short arrow) in the subcutaneous layer of the right calf and right ankle (long arrow) which is hyper-intense to adjacent muscle.



### TREATMENT

Treatment of diffuse neurofibroma is by surgical resection. The index patient is doing well 3 months post surgery. Clinical recurrences may develop even after complete excision because of the infiltrative growth pattern and the multicentricity of the tumour, therefore close follow-up of patients is essential.<sup>1-3</sup>

### OUTCOME/FOLLOW-UP/DISCUSSION

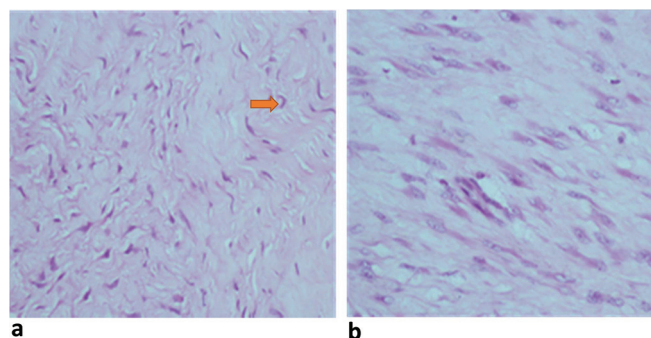
Neurofibromas are a group of common benign soft tissue tumours. They represent approximately 5% of all soft tissue tumours.<sup>4</sup> They are divided into three histological types namely localized, plexiform and diffuse.<sup>5</sup> The localized variety is the most common and most widely reported, representing approximately 90% of all neurofibroma, while the plexiform subtype is essentially pathognomonic of neurofibromatosis 1 (NF1) and also is well reported.<sup>4</sup> Diffuse neurofibroma is an uncommon form of neurofibroma typically seen in children and young adults.<sup>6,7</sup> It has been reported in the calvarium.<sup>8</sup> But it is a rare entity with a dearth in imaging literature; therefore the diagnosis may be easily missed. We present the MRI of a young adult female patient with diffuse cutaneous neurofibromatosis.

Neurofibromas are benign tumours of the peripheral nerves that develop from the proliferation of Schwann cells, perineural cells and endoneurial fibroblasts.<sup>9</sup> Diffuse neurofibroma is characterized by infiltration of skin or soft tissues. It grows between the

subcutaneous tissues without destruction of adjacent structures.<sup>7</sup> Slow growth in children with acceleration in adolescence and pregnancy is typical as seen is the patient being discussed.<sup>7,10</sup> A nerve with an ovoid or fusiform mass is the usual mode of presentation of a localized neurofibroma, while plexiform neurofibroma is serpentine in appearance and more extensive. In contrast, diffuse neurofibroma is more infiltrative with extension between tissue planes with encasement of vascular and nervous structures.<sup>10</sup> Diffuse neurofibroma is often poorly circumscribed extending along tissue planes.<sup>6</sup> Unlike the plexiform neurofibroma which is strongly associated with neurofibromatosis 1 (NF1), only an estimated 10% of patients with diffuse neurofibroma has been associated with neurofibromatosis.<sup>1-11</sup> Although earlier literature suggested diffuse neurofibromas to be most frequently found in the head and neck region, more recently Hassel *et al* reported that the trunk and extremities are the commonest locations.<sup>3,12</sup> There have been reported retroperitoneal location and associations with diffuse cystic lung disease.<sup>13</sup>

The characteristic MR imaging appearance of diffuse subcutaneous neurofibroma is iso or slightly hyperintense to skeletal muscle signal within subcutaneous tissue on  $T_1$ W MR images and mildly or markedly hyperintense to skeletal muscle signals on  $T_2$ W images. There is intense enhancement on  $T_1$ W post-contrast administration.<sup>3,4,10</sup> These findings correlate well with the MR findings in this case. On imaging with ultrasound and MR, localized neurofibromas are circumscribed soft tissue masses unlike diffuse neurofibromas that are less well-defined and more infiltrative.<sup>2</sup> Large plexiform neurofibromas are almost always seen on a background of NF1. On MRI, although plexiform neurofibroma is mass-like, it lacks internal hypointense septations demonstrated in diffuse neurofibroma.<sup>2</sup> The MR appearance and localization of diffuse neurofibroma may also be confused with those of angiomatous or fat-containing tumour, especially in a patient with a solitary lesion or one without clinical findings of neurofibromatosis.<sup>10</sup> Doppler ultrasound will reliably exclude an angiomatous mass while CT can detect fat within a tumour.<sup>14</sup> Plain radiographic findings of diffuse subcutaneous neurofibroma are not specific but can be used to exclude more aggressive lesions and closer differentials like soft tissue haemangioma where the presence of phleboliths is an important finding.<sup>6</sup>

Figure 3. Histological features of diffuse neurofibroma. Photomicrographs of surgical specimen of diffuse neurofibroma (a, b), showing spindle shaped cells with wavy to comma-shaped nuclei (arrow on photomicrograph) on a fibromyxoid background. (a = H&E  $\times 100$ , b = H&E  $\times 400$ ).



**LEARNING POINTS**

1. MRI is the imaging investigation of choice for diffuse subcutaneous neurofibroma.
2. Other imaging modalities like plain film, ultrasound and CT can be performed to rule out differential diagnosis and to define the anatomic relationships with adjacent structures.

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**CONSENT**

Written informed consent for the case to be published (including images, case history and data) was obtained from the patient(s) for publication of this case report, including accompanying images.

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