ELEPHANTIASIS NEUROMATOSA : IMAGING FINDINGS

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ABSTRACT

A 26-year-old woman with numerous Cafe-au-lait spots at the left leg since birth was presented. Plain radiographs of left leg revealed lobulated enlargement of the soft tissue of left leg from knee to foot, containing no internal calcification. Undertubulation and thickening of the cortex and the periosteum of the tibia and fibula were observed. Osteopenia with multiple extrinsic bone erosions of the tarsal bones was noted. MR imaging showed generalized thickening of the subcutaneous tissue and enlargement of the muscles of left leg, except the tibialis anterior muscle, the tibialis posterior muscle and the extensor hallucis longus muscle. Low signal intensity on T1W sequence and increased signal intensity on T2W sequence of the involved muscles were demonstrated.

INTRODUCTION

Type 1 neurofibromatosis, the peripheral form of the disease, is carried on chromosome 17.[1,2] It is a hereditary hamartomatous condition characterized by cutaneous lesions, skeletal abnormalities, and multiple nerve sheath tumors, usually neurofibromas.[1,3,4] Multiple nerve roots may be involved in a plexiform manner, being a multilobulated, tortuous entanglement or interdigitating network of tumor along a nerve and its branches.[2,3] Eventhough it is an autosomaldominant disease, about half of these cases are due to spontaneous mutation.[4]

Recent articles have described the magnetic resonance (MR) appearances of plexiform neurofibroma (PN) affecting the spine and paraspinal regions, larynx, mediastinum, and intra-abdominal organs [1,3,5,6] and one article described MR findings of PN involving the extremity.[7] This article described MR appearance of PN affecting a lower extremity with pathologic and histologic correlation.

CASE REPORT

In December 1993, a 26-year-old Thai woman presented at the surgical clinic because she had a large, lobulated-contour left leg since birth which was gradually enlarged. This enlargement was rapid and painful in the last 3 years. She still could walk and run normally. Physical examination revealed a female patient with good consiousness and good cooperation. She had numerous Cafe-au-lait spots on both anterior and posterior aspects of her trunk. The left lower extremity was enlarged and lobulated without varus or valgus deformity. The lesion involved the entire left lower leg and foot. Other physical examination was unremarkable. The family history was obtained and disclosed that her

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three first-degree relatives and other three second-degree relatives had Cafe-au-lait spots, but to a lesser extent. Plain radiographs of left leg revealed lobulated enlargement of the soft tissue of the left leg from knee to foot without internal calcification. Undertubulation and cortical/ periosteal thickening of the tibia and fibula was shown. Osteopenia and multiple varied-size extrinsic bone erosions were noted at the tarsal bones (Fig.1). MR imaging of left leg (Fig.2,3) demonstrated generalized thickening of the subcutaneous tissue and enlargement of the muscles of left leg, except of the tibialis anterior muscle, tibialis posterior muscle, and extensor hallucis longus muscle. The signal intensity (SI) of the involved muscles were abnormal with less SI than normal muscle on T1W images (T1WI) and increased SI on T2WI and gradient recalled echo images (GREI). The exceptional muscle groups showed normal size and signal intensity. Marrow compartment appeared normal. Bone erosions at the tarsal bones on plain radiographs were produced by the soft tissue mass. The diagnosis was compatible with neurofibromatosis. No scoliosis or intrathoracic lesion seen on chest film.

The patient was undergone contouring procedure, and the excisional biopsy was taken from the left foot. The specimen consisted of elongated elliptical shaped skin with underlying mass, measuring 25 X 5 X 4 cm. The epidermis was brown and wrinkle. The underlying mass showed shiny white, soft appearance with an infiltrative border into the adjacent dermis and subcutaneous adipose tissue. Microscopically, the mass was composed of diffuse proliferation of compactly arranged spindle cells with scanty but extensively elongated cytoplasm which were lightly eosinophilic and fibrillary (Fig. 4a). Their nuclei were wavy or comma-shaped (Fig. 4b). Several expanded nerves were seen as the result of infiltration by the neoplasm. Dense eosinophilic rim representing the remnant of the original nerve was also present and it appeared as a small nodule at the low-power field. The diagnosis of plexiform neurofibroma was given. All surgical margins were not free. The patient had necrotic skin over the posterior ankle post operatively and was treated medically. She had an appointment for the second surgery in June 1994, but lost follow up.

DISCUSSION

Peripheral nerve tumors in neurofibromatosis most frequently involve neural supporting tissue. The two most common benign forms are solitary or multiple neurofibromas and neurilemmomas (schwannoma). The latter are encapsulated lesions that lie on the surface of the nerve. They are more common than neurofi-bromas, which infiltrate involved nerves diffusely and incorporate all nerve elements (i.e. Schwann cells, nerve fibers, and fibroblasts). Tumors of nerves may appear as focal fusiform enlargement or diffuse, multinodular, or coalescent sheetlike masses.[4,8] Benign plexiform or cirsoid neurofibromas, observed only in neurofibromatosis, appear as bizarre soft tissue networks that interdigitate imperceptibly with adjacent fat and muscle, recur after resection,[3,6] and have a potential for malignant degeneration. They are recognized most easily in massively enlarged extremities (i.e. elephantiasis neuromatosa), although the face, head and trunk also may be involved. Such lesions may occur anywhere along the course of central or peripheral nerves,[4] or within deep or superficial tissues.

The MR signal characteristics of PN has been described to be isointense or slightly greater than muscle on T1WI, and markedly increased on T2WI.[1,3,5-7] Previously, the low attenuation of neurofibromas on CT has been attributed to increased amount of endoneural myxoid matrix .[1,5,9] This matrix contains acid mucopoly-saccharides and a large amount of tissue fluid. The residual axons are spread by the matrix to produce the fusiform shape or the neurofibroma.[10] The lengthened T2 of the neurofibroma is probably due

to increased water content of the matrix. The slightly greater SI of the neurofibroma on T1WI can be attributed to relative shortening of T1 by the interaction of the large mucopolysaccharides molecules with the tissue water. Large protein molecules have been demonstrated to slow the tumbling rates of water molecules resulting in accelerated relaxation rates.[5,11]

Our case with elephantiasis neuromatosa exhibited slightly lower signal intensity than muscle on T1WI which could be attributed to cystic degeneration,[1] and to the increased fluid content within the lesion. The myxoid matrix found in histological examination as well as the high fluid content may account for the increased SI on T2WI. We did not find the central area of decreased SI on T2WI described in paraspinal lesions[5] in our case. The lesion in our study had infiltrative margins that blended imperceptibly with adjacent fat and muscles but still showed benignity histologically; this finding corresponds to what were described by other authors.[6] The low-SI linear or serpentine structures within the lesion, obviously seen on T2WI and GREI, representing separated muscle bundles. The dysplastic change of the tibia and fibula included undertubulation and thickening / hypertrophy of the cortex. The latter occured partly as result of the incooperation of the overgrowing periosteum which was a reactive change to the adjacent soft tissue tumor. The extrinsic erosive change of the tarsal bones reflected the aggressive pattern of the tumor growth.

In conclusion, we report imaging findings of a case of elephantiasis neuromatosa of left lower extremity which showed less SI than muscle on T1WI and high SI on T2WI. The selective involvement of particular muscle groups and preserved other groups characterized the plexiform manner of involvement along the course of peripheral nerves.

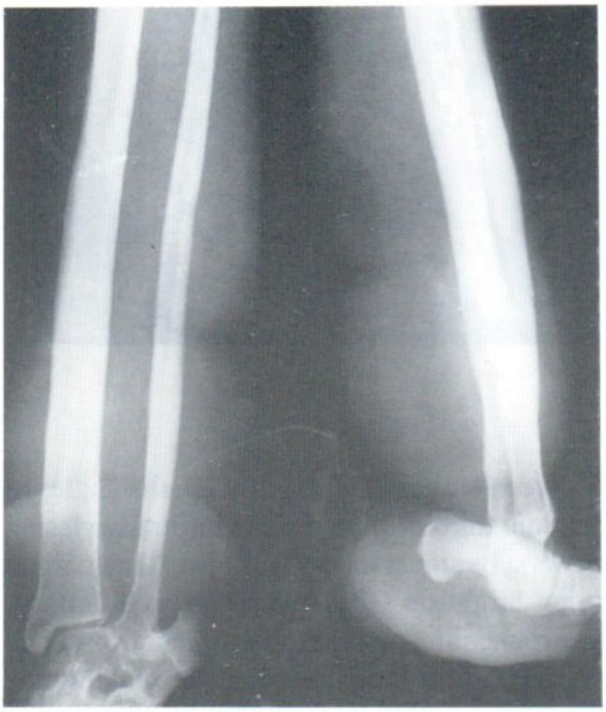


Fig. 1. Plain radiograph of left leg reveals multi-lobulated enlargement of soft tissue as well as skeletal abnormalities onsisting of cortical thickening and hypertrophy which are partly incorporated by the overgrowing periosteum.

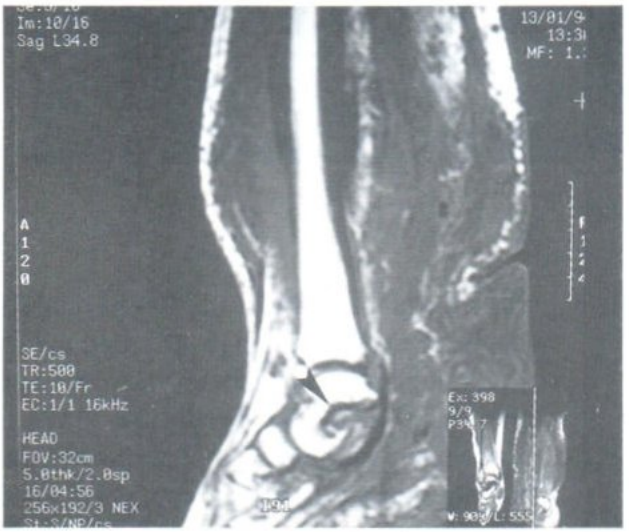
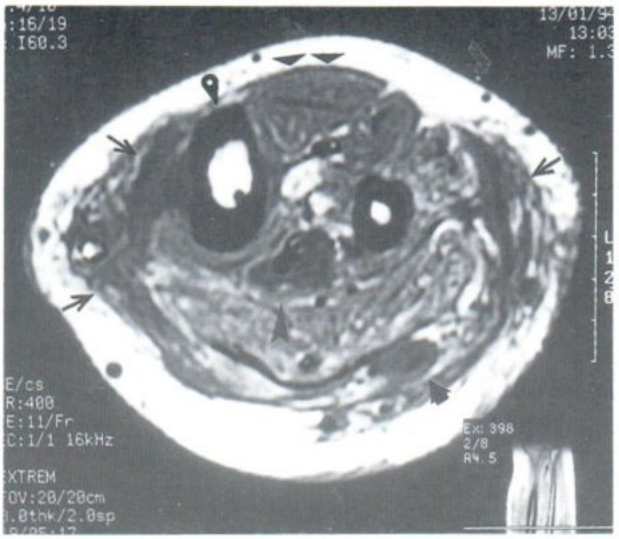
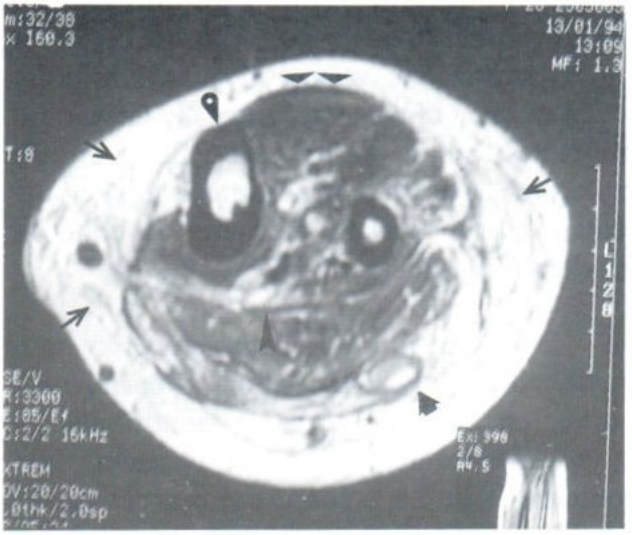


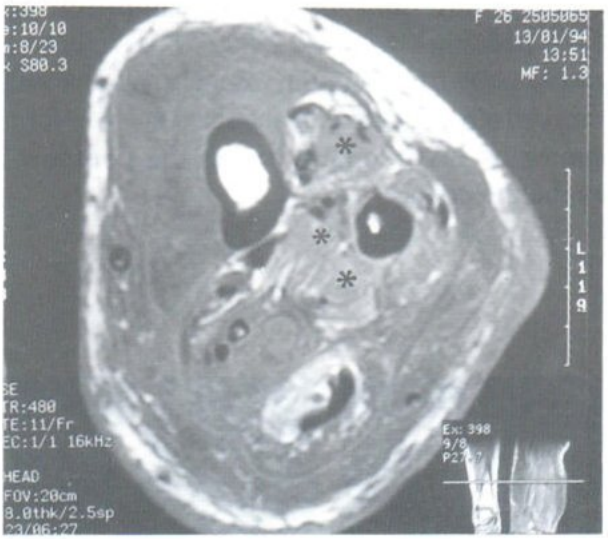
Fig. 2. T1- weighted (500/10 [TR/TE]) sagittal MR image reveals an infiltrating soft tissue PN showing lower-than-muscle SI and causing bone erosion of the tarsal bones (black arrowhead).



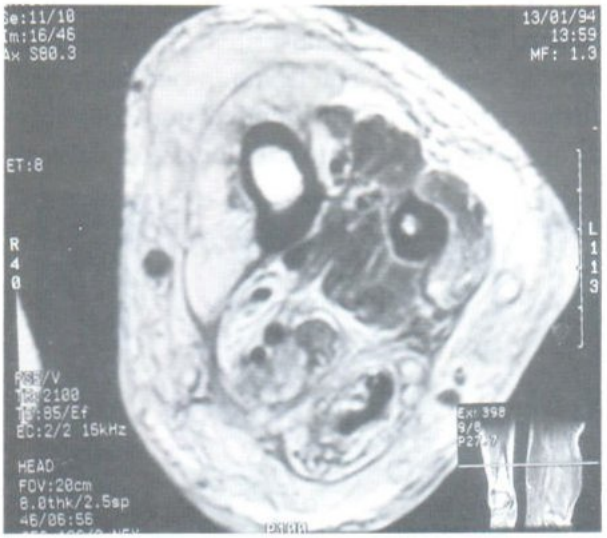
3(a)



3(b)



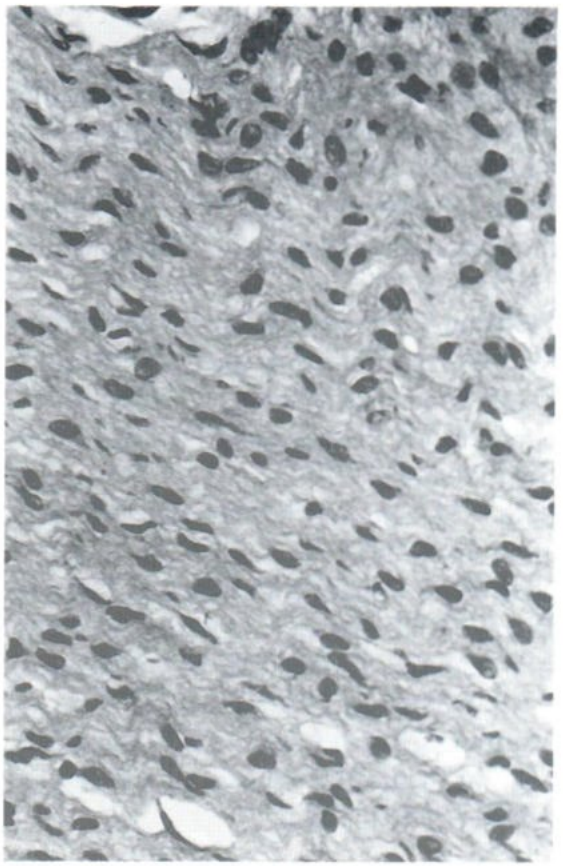
3(c)



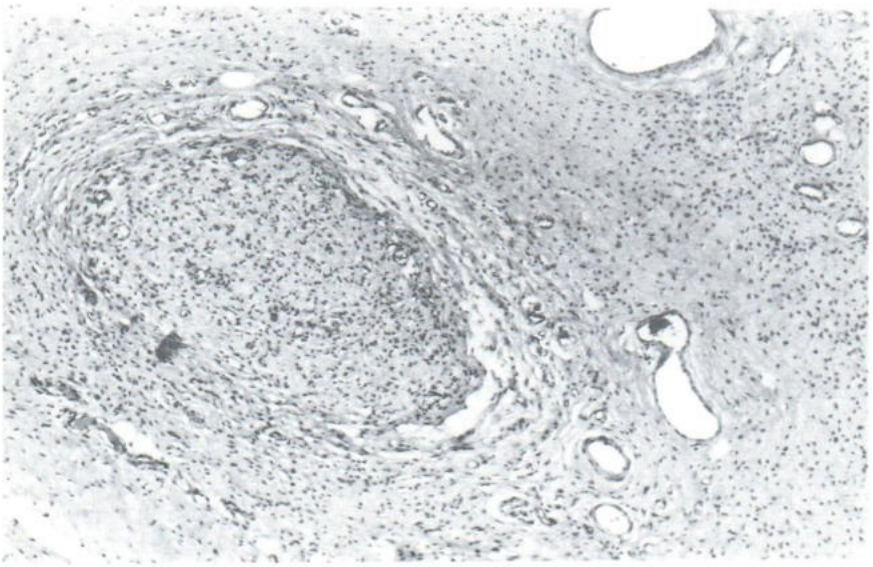
3(d)

Fig.3 **(a)** T1-weighted (400/11 [TR/TE]) and **(b)** gradient recalled echo (GRE)(600/20; flip angle, 15 degrees) axial MR images through the level of middle third of tibia reveals infiltrative lesion involving both subcutaneous fat and muscles (arrows). Its signal intensity is lower than normal muscles (broad arrowheads) on T1W1 and turning bright on GRE image. A well-defined, focal neurofibroma is observed within lateral gastrocnemius muscle (short thick arrow) and having internal cystic component. Another focal plexiform lesion is noted surrounding the posterior tibial neurovascular bundle (arrowhead). Abnormal thickening of bony cortex is also observed (round arrowhead.) **(c)** T1- weighted (480/11) and **(d)** GRE (680/20; flip angle, 15 degrees) axial MR images 20 mm below a) and b) exhibit more extensive lesion. On T1WI (c) the signal intensity of the lesion is lower than normal muscles (asterisks). The low-SI linear or serpentine structure seen within the lesions on GRE image represent muscle bundles and fascia separated by the tumor.

4(a)



4(b)



4(c)

Fig. 4. **(a)** (x40) The plexiform neurofibroma involves the dermis. Its border was well-defined but unencapsulated. The mass composed of spindle cells. **(b)** (x200) The cellular area of the plexiform neurofibroma shows proliferation of wavy, fibrillary elongated spindle cells. The nuclei are oval, wavy or comma-shaped. **(c)** (x40) One of a number of expanded nerves in plexiform neurofibroma. Its perineurium was thickened and extended into the adjacent tissue.

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