
CASE REPORT

Fibrolipomatous Hamartoma of Tibial Nerve and Its Musculoskeletal Associations

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ABSTRACT

Fibrolipomatous hamartoma is a rare condition caused by hypertrophy of mature fat and fibroblasts in the epineurium, resulting in fibrofatty infiltration and an enlarged peripheral nerve. The median nerve is most commonly affected, in which case it can be associated with macrodactyly and macrodystrophia lipomatosa, characterised by nerve territory-oriented macrodactyly as well as increased fat deposition in the subcutaneous tissue and soft tissues. Our objective was to describe the pathognomonic magnetic resonance imaging findings of this condition in an uncommon location, namely the tibial nerve, and highlight its associated musculoskeletal anomalies, namely macrodactyly, polydactyly, and syndactyly.

Key Words: Hamartoma; Lipomatosis; Macrodactyly of the foot; Peripheral nervous system diseases; Tibial nerve

中文摘要

脛神經纖維脂肪瘤性錯構瘤及其相關的骨骼肌肉異常

謝健榮、黎國忠、趙朗峰、黎國鴻、陳文光

纖維脂肪瘤性錯構瘤是指在神經外膜上成熟脂肪細胞和纖維母細胞增生的一種罕見情況，形成纖維脂肪組織浸潤神經外膜，並導致受累的周圍神經增粗。最普遍受影響的是正中神經，受累神經可併發巨指和脂肪瘤性營養異常性巨大發育。常見情況包括病人出現受累神經分布區域的巨指，以及皮下組織和軟組織有大量脂肪積聚。本文描述的病例出現在罕見的脛神經位置，我們探討其特殊的磁共振影像及相關的骨骼肌肉異常，即巨指、多指和併指。

INTRODUCTION

Fibrolipomatous hamartoma, first described by Mason in 1953,¹ is a rare condition characterised histologically by fibrofatty infiltration within the nerve, associated with perineural and endoneural fibrosis, and thickening of nerve fascicles. Fusiform enlargement of the affected nerve and fatty infiltration between the thickened nerve

fascicles are its key imaging findings. It most commonly occurs in the median nerve. Lower limb involvement is rarely reported in the literature. The purpose of this report was to describe a patient with a fibrolipomatous hamartoma of the tibial nerve, a less commonly encountered site, as well as its associated bone and soft-tissue anomalies.

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CASE REPORT

A 54-year-old woman was referred to our department for investigation of local gigantism of the right foot in 2006. She noticed enlargement of her right foot, particularly the third and fourth toes, since childhood. She also found a painless soft-tissue mass at the medial

aspect of her right ankle for more than 10 years, which remained static in size. Despite the local gigantism of her right foot, she remained relatively asymptomatic with only mild pain over her sole from plantar keratosis. Physical examination revealed diffuse enlargement of her right foot, with exceptionally large third and fourth toes. No dilated superficial vein or vascular mark was noticed. The right ankle mass was non-tender and non-compressible.

Magnetic resonance imaging of the right lower leg, ankle, and foot was performed using a 1.5-Tesla Siemens Magnetom Avanto machine. The imaging protocol included TSE-T1-TRA, TSE-T2-FS-TRA, STIR-T2-COR, TSE-T1-GAD-FS-COR, TSE-T1-GAD-FS-TRA, and TSE-T1-GAD-FS-SAG.

The lower part of the right tibial nerve demonstrated marked fusiform enlargement, associated with abundant T1-hyperintense tissue with signal suppression on fat-saturated sequences, suggestive of lipomatous tissue interspersed among the thickened hypointense nerve fascicles. The lesion showed no contrast enhancement

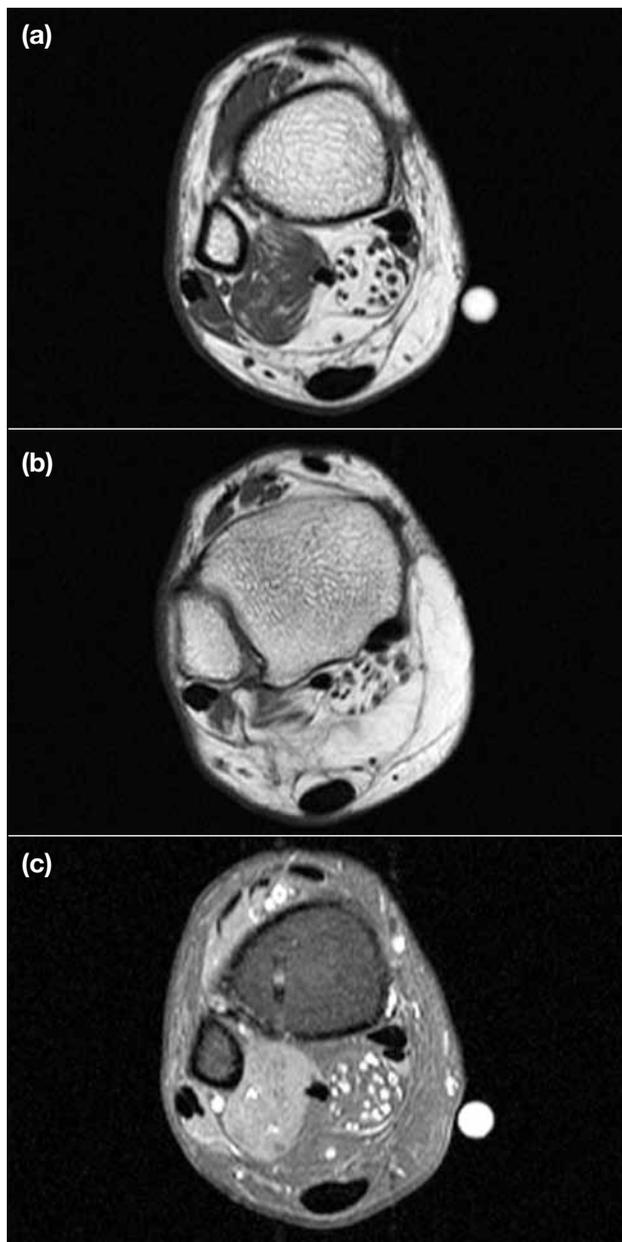


Figure 1. Axial T1-weighted images showing marked enlargement of right tibial nerve, at the level of (a) distal calf and (b) tibial plafond. Oil capsule has been placed in (a). Multiple hypointense dots, representing the thickened nerve fascicles, are surrounded by the T1-hyperintense fatty tissue. The lesion has the characteristic co-axial cable-like appearance on this axial image. (c) A post-gadolinium T1-weighted image (at the level of a) shows no contrast enhancement of the lesion.

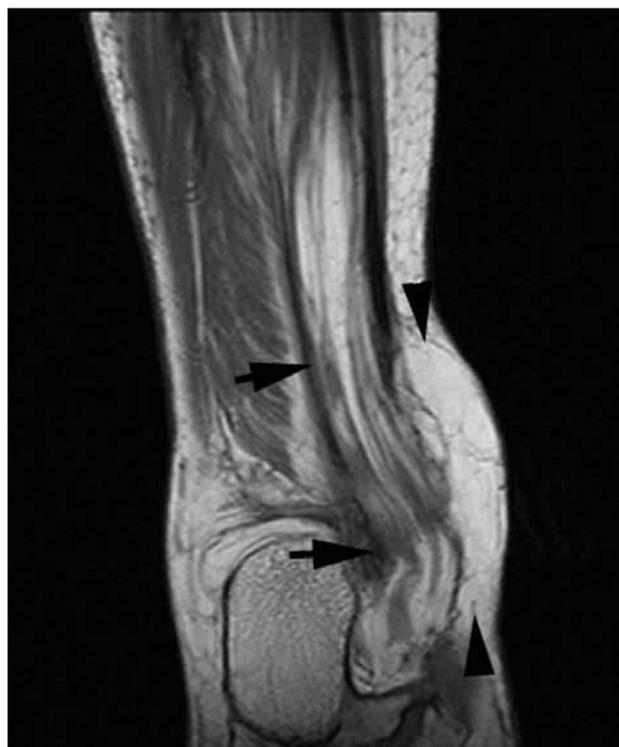


Figure 2. A coronal T1-weighted image demonstrates the deposition of fatty tissue which infiltrates between the linear T1-hypointense nerve fascicles of the right tibial nerve (arrows), with a characteristic spaghetti-like appearance. Increased deposition of subcutaneous fat is seen at the medial aspect of right ankle (arrowheads).

after gadolinium administration (Figure 1). The lipomatous lesion involved the lower part of the tibial nerve, and extended distally to the proximal portion of the lateral and medial plantar nerves. These features were compatible with fibrolipomatous hamartoma of the tibial nerve.

Increased deposition of the subcutaneous fat was noted at the medial and posterior aspects of the ankle, posterior part of the sole, with disproportionately enlarged third and fourth toes (Figures 2 and 3). Streaky T1-hypointense and T2-hypointense signal intensities suggestive of fibrous strands were observed inside the prominent subcutaneous fat. The features were compatible with macrodystrophia lipomatosa and macrodactyly associated with fibrolipomatous hamartoma.

Associated skeletal anomalies were also found. The plain radiographs correlated with the appearance of the enlarged and deformed phalanges of the third and fourth toes (Figure 4). New bone formation around the metatarsals and phalanges of these two digits appeared to have caused fusion of the digits. In addition, a bi-



Figure 3. A sagittal T1-weighted image illustrates the associated musculoskeletal anomalies of fibrolipomatous hamartoma. The spaghetti-like appearance of fatty infiltration within the thickened right tibial nerve (large arrowhead) is again shown. A bi-phalangeal structure is noted arising from the undersurface of the calcaneum (arrows). Increased subcutaneous fat is noted in the enlarged digit (small arrowheads) and at the posterior aspect of right ankle compatible with macrodactyly and macrodystrophia lipomatosa.



Figure 4. (a) Oblique and (b) lateral radiographs of the right foot shows diffuse soft tissue and bone overgrowth, and deformity of the third and fourth digits. Prominent soft-tissue overgrowth over the sole, particularly the heel, is also observed. An anomalous bi-phalangeal structure is noted, articulating with the medio-inferior aspect of the calcaneum.

phalangeal structure arising from the undersurface of the calcaneum, representing a form of polydactyly, was noticed (Figure 4).

The patient opted for conservative treatment. She could walk for a few hours with special shoes without significant symptoms or discomfort, and currently undergoes regular follow-up at our orthopaedic clinics.

DISCUSSION

Fibrolipomatous hamartoma is a rare condition, the exact incidence of which has not been well-described in the literature. It typically presents in childhood or early adulthood. It has also been called fibrolipomatous nerve enlargement, lipofibroma, fibrofatty overgrowth, neural fibrolipoma and neurolipoma. In 2002, the World Health Organization designated it as a lipomatosis of the nerve.²

Adipose tissue is normally found in the peripheral nerves, within the perineurium and epineurium. In fibrolipomatous hamartoma, the perineural and epineural compartments of the involved nerve are infiltrated by mature adipose tissue admixed with fibrous tissue, which dissect between and separate individual nerve bundles. Atrophy with concentric perineural fibrosis causing thickening of the nerve fascicles is also present. The affected nerve may also show other changes such as perineural septations, microfascicle formation, and pseudo-onion bulb formation.^{2,3} The exact pathogenesis of fibrolipomatous hamartoma is not yet well understood. It is not associated with any syndrome or hereditary predisposition. It has been postulated to be congenital in nature, as well as acquired after trauma, nerve irritation, and inflammation.⁴

Patients with fibrolipomatous hamartoma typically present before the age of 30 years, more commonly in childhood; symptomatically they have a slowly growing soft-tissue mass, followed by pain, numbness, and decreased sensory and motor function. The most frequent initial clinical feature is a painless swelling or mass. Neurological deficits arising from dysfunction or compression of the nerve are usually late symptoms, which usually occur several years after onset of the condition.⁴ They include carpal tunnel syndrome from median nerve involvement, due to constraints of the flexor retinaculum.^{5,6} The upper extremity is more often involved than other sites (in about 78 to 96% of cases); there being a marked predilection for median nerve, but the ulnar nerve,^{7,8} radial nerve,⁹ and

brachial plexus are other reported sites. In about 4 to 22% of the instances, the condition occurs in the lower extremity; case reports and series have described sciatic,^{8,10} superficial peroneal,^{11,12} plantar³ and tibial nerve involvement.¹¹

Findings from imaging are classical and pathognomonic. Ultrasonography usually shows a hyperechoic mass containing co-axial hypoechoic bands, corresponding to the thickened nerve fascicles. Computed tomography demonstrates fusiform nerve enlargement with fatty tissue interdigitating between serpentine soft-tissue structures. On axial images, magnetic resonance imaging reveals T1-hyperintense and T2-hyperintense tissue infiltrating between the hypointense dots (about 2-3 mm thick), and has a co-axial cable-like appearance; on coronal images the serpentine linear hypointense structures show a surrounding spaghetti-like appearance.^{8,11,13,14} The fat signal intensity and characteristic cable-like or spaghetti-like appearance of fibrolipomatous hamartoma are pathognomonic of this entity (Figures 1 and 2). Thin hypointense septations are also found within the fat separating the nerve fascicles, which presumably represents thickened perineurium.¹¹ No enhancement is identified on post-gadolinium fat-saturated sequences.⁵ Fibrolipomatous hamartoma differs from its diagnostic mimics. For example, a well-encapsulated intraneural lipoma arises from the perineurium and presents as a focal fatty mass separated from and eccentric to the nerve fascicles.¹⁵ The presence of fatty infiltration also differentiates fibrolipomatous hamartoma from other causes of nerve enlargement, e.g. schwannoma, post-traumatic neuroma and chronic inflammatory demyelinating polyneuropathy. The presence of contiguous involvement of the tibial nerve and distally the plantar nerves also illustrates another important growth pattern of fibrolipomatous hamartoma, namely extension of the lesion into distal nerve branches.^{6,11} Due to the pathognomonic diagnostic features of this condition, the need for biopsy is usually obviated.⁸

This condition is often associated with macrodactyly (in 27 to 67% of affected persons), and is due to soft-tissue and osseous overgrowth, known as macrodystrophia lipomatosa. The latter condition shows a female preponderance, and is caused by bony overgrowth and fibrofatty tissue deposition in subcutaneous tissues, tendons, and muscles (often in the territory supplied by the affected nerve).^{12,13,16} Osseous overgrowth is more marked volarly and distally, and is commonly

associated with premature osteoarthritis. Due to osseous and fibrofatty tissue overgrowth, the phalanges are usually enlarged in all dimensions. Thus, macrodactyly is an essential feature of macrodystrophia lipomatosa. The latter must be differentiated from the isolated fat deposition in the muscle without macrodactyly, which can result from nerve malfunction / denervation.¹¹ As in our case, macrodactyly and deposition of fibrofatty tissue (macrodystrophia lipomatosa) in the subcutaneous region facilitates the diagnosis of fibrolipomatous hamartoma.

Other less common skeletal and soft-tissue anomalies have been associated with this condition. Early maturation of epiphyseal ossification centres of the phalanges and metatarsals, syndactyly, polydactyly, brachydactyly, and symphalangism have all been encountered in patients with macrodystrophia lipomatosa,^{17,18} as illustrated by the polydactyly in our case. Additional associations with fibrolipomatous hamartoma include exostosis, soft-tissue lipomas, vascular malformations, and soft-tissue calcifications.¹⁹ Thus, a search for other soft-tissue lesions and osseous deformities is warranted in all affected patients.

The treatment of the fibrolipomatous hamartoma remains controversial. Surgery is not recommended in all cases, due to detrimental effects on motor and sensory functions, apart from having the potential to cause postoperative neurogenic pain. Moreover, in this infiltrative process, the optimal resection margin can be difficult to achieve. Therefore conservative treatment coupled with decompression of compromised nerves is generally adopted.⁴

In summary, we report a case of fibrolipomatous hamartoma of the tibial nerve, which is a less commonly involved site. This condition is characterised by fusiform nerve enlargement with fatty tissue infiltrating between the thickened nerve fascicles. Associated macrodystrophia lipomatosa and macrodactyly, together with other skeletal anomalies namely polydactyly, were observed in our patient.

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