

Lipofibromatous Hamartoma of a Digital Nerve

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Lipofibromatous hamartoma of a nerve is a gross enlargement of the nerve caused by large proportions of fat and fibrous tissue, with epineural and perineural proliferation.¹⁻⁵ More than 90 cases have been described in the English-language literature.^{1,5} Although most commonly reported in the median and digital nerves, the lesion also occurs in the radial,^{6,7} ulnar,^{8,9} sciatic,¹⁰ cranial,¹¹ and plantar¹² nerves. The etiology of this benign tumor is unknown but is considered by some authors to be congenital.^{1,2,12}

The mass is usually slow-growing and may cause nerve compression resulting in sensory change, pain, and loss of motor function.¹⁻¹² Patients usually present before adulthood. Biopsy has been needed to confirm the diagnosis and continues to be the standard approach to establish the diagnosis; however, recent reports suggest that the magnetic resonance imaging (MRI) appearance of this entity is pathognomonic.^{4,13}

Treatment of lipofibromatous hamartoma is controversial. Recommendations include decompression of the carpal tunnel (when the lesion involves the median nerve at the wrist), decompression and debulking of the fibrofatty sheath, microsurgical dissection of the neural elements, excision of the involved nerve segment with or without nerve grafting, and observation for asymptomatic patients.¹⁻¹⁵

We present the case of a manual laborer with lipofibromatous hamartoma of a digital nerve with normal sensory and motor function before surgery but with an enlarging first web space mass altering the dexterity and functional use of the hand. The patient was treated with incisional biopsy and debulking of the tumor to improve first web space depth and digital range of motion. After presenting this case, we review the literature.

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CASE REPORT

A right-hand-dominant man in his mid-20s presented to our hand clinic for evaluation of a left thumb mass. For 8 years, he had had a mass along the ulnar aspect of the left thumb. He denied pain, sensory changes, and cold intolerance, but over 18 months the mass had gradually increased in size, and gripping and grasping activities at work had become difficult because of loss of depth in the first web space and diminished thumb apposition. The patient did not recall a history of trauma to the hand, and his medical history was unremarkable. There was no family history of neurofibromatosis or macrodactyly.

Physical examination revealed a soft, mobile, non-tender deep mass (length, 10 mm; width, 5 mm) overlying the ulnar aspect of the proximal phalanx of the thumb extending into the first web space. There was a 30° loss of thumb flexion compared with the contralateral side, but the patient had full thumb interphalangeal extension. Sensation was intact to light touch. Two-point discrimination was 5 mm in the ulnar digital nerve distribution of the thumb and normal in the rest of the hand. There was no Tinel's sign, and there was no pulse palpated in the mass.

Plain radiographs showed normal anatomy with no evidence of bony abnormality.

The differential diagnosis included lipoma, liposarcoma, synovial cell sarcoma, nerve sheath tumor, macrodystrophia lipomatosis, neurofibromatosis, and lipofibromatous hamartoma.

The patient began having difficulty doing his work and wanted the mass excised. As restricted thumb flexion interfered with the man's work, we felt that surgical exploration and tumor debulking of the left thumb were indicated. The likelihood that this mass represented a malignancy was



Figure 1. Intraoperative photograph shows lipofibromatous hamartoma of the ulnar digital nerve of the thumb.



Figure 2. Intraoperative photograph shows that the continuity of the ulnar digital nerve was preserved after debulking of the lipofibromatous hamartoma of the nerve.

extremely low, based on the chronicity, size, and location of the mass; however, before surgery, we planned an intraoperative frozen section and told the patient that additional surgery and possible ray resection might be needed if the planned intraoperative pathology revealed a malignancy. A midlateral incision along the ulnar border of the thumb extending to the thenar crease was used to expose the mass and to identify the digital nerves to the left thumb and index finger. This approach allowed for direct access to the mass and for potential ray amputation in the event that the mass was found to be malignant. A 60×10×10-mm yellow-orange lipomatous mass, encompassing the ulnar digital nerve of the thumb proximally at its origin in the palm and distally to the level of interphalangeal joint, was found (Figure 1). The radial digital nerves to the thumb and index finger were normal. Intraoperative frozen sections revealed fibroadipose tissue infiltrating and distorting the nerve fascicles, consistent with lipofibromatous hamartoma of the nerve. The tumor was then carefully dissected and debulked. The continuity of the digital nerve was maintained (Figure 2). The specimen was sent for final pathologic study, which confirmed the intraoperative diagnosis of lipofibromatous hamartoma. A microphotograph from the surgical specimens is shown in Figure 3.

After surgery, the patient continued to have full motor and sensation to light touch. He returned to work 3 weeks after surgery and achieved full thumb flexion compared with the contralateral side. Six months after surgery, 2-point discrimination remained unchanged from presurgical status, and the patient had no restrictions at work. There was no evidence of recurrence of the tumor at 1-year follow-up.

DISCUSSION

Four types of lipomatous masses associated with peripheral nerves have been identified: soft-tissue lipoma, intraneural lipoma, macrodystrophia lipomatosa, and lipofibromatous hamartoma.¹⁶ Soft-tissue lipomas are true benign neoplasms originating from adipose cells, which can cause peripheral nerve compression extrinsically.^{17,18} Intraneural lipomas arise from adipose cells within the nerve. These benign tumors are encapsulated, clearly defined, and easily excised.^{19,20} Macrodystrophia lipomatosa is characterized by diffuse enlargement of the digits,

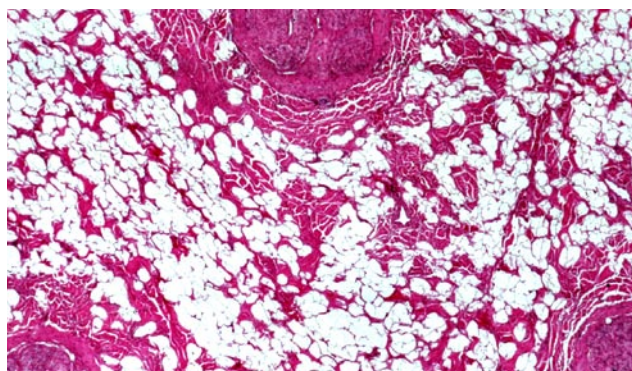


Figure 3. Microphotograph of the debulked lipofibromatous hamartoma of the ulnar digital nerve of the thumb shows fibroadipose tissue infiltrating nerve fascicles (hematoxylin-eosin, original magnification ×100).

usually the thumb and index finger, caused by fatty infiltration and hypertrophy of all components of the digit, including skin, bone, and nerves.¹⁶ Lipofibromatous hamartoma, first described in the English-language literature by Mason²¹ in 1953, is a rare condition characterized by diffuse enlargement of a nerve trunk, caused by overgrowth and fibrofatty tissue proliferation within the epineurium, perineurium, and endoneurium without overgrowth of the surrounding tissues.^{3,4} The individual nerve fibers are normal by microscopy.^{4,14} The lesion is described as a hamartoma because fat and fibrous tissues are normal constituents of the connective tissue stroma of nerve. In this case report, all criteria are satisfied for diagnosing lipofibromatous hamartoma of the digital nerve.

The etiology of lipofibromatous hamartoma is unknown. Genetic factors may play a role,^{1,4} though no specific genes have been identified as contributing to this benign perineural fibrofatty overgrowth. The disease most commonly involves the median nerve and its distribution. Of 43 lesions in 4 large series, 35 were in the median nerve, 3 in the ulnar nerve, and 5 in the sciatic nerve.^{1,2,5,13} This striking predilection for the median nerve remains unexplained. It is the most common condition associated with macrodactyly of the hand and seems to be more common in women than in men. However, lipofibromatous hamartoma without macrodactyly has an equal sex distribution.^{2,22} Other associated conditions are bony exostoses,^{14,23} ectopic calcification within median nerve and soft tissue,^{2,14,24} and Klippel-Trénaunay syndrome (gigantism with multiple vascular malformation).² Investigation of these associated conditions may help us delineate the pathophysiology of this rare disease.^{1,2}

Results from previous studies suggest that the MRI appearance of a lipofibromatous hamartoma is pathognomonic (Figure 4).^{4,13} The nerve is abnormally enlarged, with the fascicles appearing as a coaxial cable in low-signal intensity on both T₁- and T₂-weighted axial images; the fascicles are separated from one another by uniformly dispersed fat, which appears on MRI as high-signal intensity on T₁-weighted images and as low-signal intensity on fat-suppressed T₂-weighted images, consistent with benign lipomatous tissue as subcutaneous fat in other locations. On

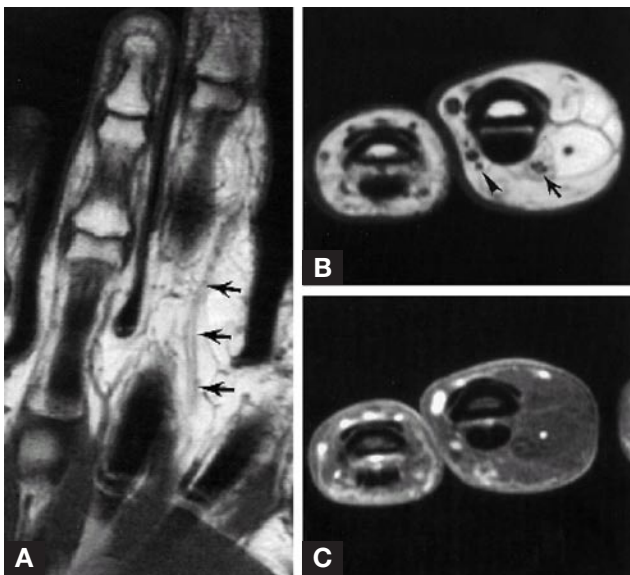


Figure 4. Magnetic resonance imaging scans of a lipofibromatous hamartoma of the long finger: (A) Coronal T_1 -weighted image shows radial digital nerve (arrows) surrounded by high-signal intensity fat (TR/TE, 600/12 ms). (B) Axial T_1 -weighted image (TR/TE, 600/12 ms). Note the normal-appearing ulnar digital nerve (arrow head) compared with the 4 radial digital nerve fascicles (arrow) separated by high-signal fat. (C) Axial T_1 -weighted fat suppression sequence after administration of intravenous gadolinium diethylenetriaminepentaacetic acid shows that the digital nerves remained low signal (TR/TE, 600/12 ms). Unlike the vessels, which are high-intensity signal, the mass did not enhance, and the fat within the lesion looks similar to normal subcutaneous fat irrespective of the imaging sequence. Reproduced with permission from Leo BM, Chhabra A, Kaplan PA, Degnan GD. A long finger mass in a 43-year-old woman. *Clin Orthop.* 2003;(408):344-351.

coronal section, the nerve has a spaghetti or strands-of-hair appearance, again in low-signal intensity and embedded in fatty tissue. These characteristics of lipofibromatous hamartoma on MRI differ from those of virtually all other nerve lesions associated with focal or diffuse nerve enlargement, including intraneural lipoma, ganglion cyst, traumatic neuroma, plexiform neurofibroma, and hereditary hypertrophic interstitial neuritis of Dejerine-Sottas disease, all of which show high-signal intensity on T_2 -weighted images (which is lacking in lipofibromatous hamartoma). Therefore, when fat is evenly distributed among nerve fascicles in conjunction with low-signal intensity on T_2 -weighted images, the diagnosis of lipofibromatous hamartoma can be made.^{4,13,25-27}

Treatment of lipofibromatous hamartoma is controversial because the lesion cannot be completely excised without sacrificing the involved nerve. However, the benign tumor can be significantly debulked to improve functional outcomes. Whereas some authors have reported that excision of the involved nerve did not lead to apparent detriment to sensibility,^{10,15} others have reported neural deficit.²⁴ Therefore, excision of the nerve is not recommended. Extensive intraneural dissection may lead to

neurologic deficit caused by ischemic complications.² In this case report, however, lesion debulking did not effect neurologic changes. Given the existing knowledge, we recommend that each patient be treated individually, according to clinical presentation and diagnosis. Surgical decompression and debulking are recommended for patients with motor or sensory deficits, such as carpal tunnel syndrome and restricted digit range of motion. In an asymptomatic patient who has been diagnosed on the basis of clinical history and MRI studies, a conservative approach (eg, observation) is appropriate.^{1-3,5,14}

AUTHORS' DISCLOSURE STATEMENT

The authors report no actual or potential conflict of interest in relation to this article.

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This paper will be judged for the Resident Writer's Award.