# Fibrolipomatous hamartoma of the nerve: A clinicopathologic report of 13 cases

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***Background:*** Fibrolipomatous hamartoma of the nerve is a rare benign infiltrating condition of peripheral nerves with prominent cutaneous findings that has not being well described in the dermatologic and dermatopathologic literature.

***Objective:*** We sought to evaluate the clinical and histopathological features of this rare condition.

***Methods:*** We reviewed the clinicopathologic features of 13 cases to delineate their clinical presentation and histopathologic spectrum.

***Results:*** All patients presented with unilateral lesions on the thenar areas, fingers, or both. In 7 cases the lesions presented congenitally and in 6 cases the lesions presented sporadically. Histologically, we found 2 patterns that have only been rarely mentioned before including cases with intraneural perineurioma-like features and cases with marked nerve hyperplasia.

***Limitations:*** Only 13 cases were included in our study.

***Conclusions:*** This condition is an uncommon entity. The diagnosis of this disorder can be highly suspected on its macroscopic features. Predilection of the median nerve and the frequent association with macrodactyly are characteristic clinical findings. (J Am Acad Dermatol 2014;70:736-42.)

***Key words:*** fibrolipomatous hamartoma of the nerve; macrodactyly; median nerve; Proteus syndrome; syndactylia.

Fibrolipomatous hamartomas of the nerve are rare, benign, fibrofatty malformations of peripheral nerves. Other terms applied to this condition are "perineurial lipoma," "intraneural lipoma," and "lipomatous hamartoma."[1-3] Most cases occur in the first 3 decades of life (often at birth or early childhood) and it is believed to be of congenital origin.[3] Most cases present as isolated lesions but one third are associated with macrodactyly, which is referred to as macrodystrophia lipomatosa.[4] More than 80% of fibrolipomatous hamartomas arise exclusively in the median nerve; however, they can also occur in a digital nerve alone.[5,6]

Herein, we described 13 cases of fibrolipomatous hamartomas of the nerve in which cutaneous symptoms were prominent, highlighting their salient clinical and histopathological features. Despite its tendency to occur on the hands, and having characteristic cutaneous changes, they have only rarely been reported in the dermatologic literature.[7]

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## METHODS

Thirteen cases of fibrolipomatous hamartomas of the nerve form the basis of our study. The cases included 7 female and 6 male patients, aged from 7 to 52 years (mean: 29.5 years). Clinical data and follow-up information were obtained from the hospital records. All cases were seen in consultation at the Medical College of Wisconsin, Milwaukee, Wisconsin. The study was approved by the Medical College of Wisconsin Institutional Review Board Committees.

All tissues were fixed in neutral buffered formalin and embedded in paraffin for histologic processing. Tissue sections were stained with hematoxylin-eosin for conventional histology. Representative paraffin-embedded tissue blocks were cut and processed for immunohistochemical studies. Each case was analyzed with 1 to 5 histologic sections stained with hematoxylin-eosin. Immunohistochemical staining was performed in an autostainer (DAKO Corp, Carpinteria, CA) using a standard avidin-biotin peroxidase complex technique. Heat-induced epitope retrieval was applied as pretreatment for selected markers. Diaminobenzidine was used as chromogen with light hematoxylin as counterstain. The antibodies used included: epithelial membrane antigen (EMA) (DAKO, E29, 1:450) and S100 (DAKO, 4c4.9, 1:75). Appropriate positive and negative controls were run concurrently for all the markers tested.

**CAPSULE SUMMARY**

- Fibrolipomatous hamartoma of the nerve is a rare benign condition of peripheral nerves that shows characteristic cutaneous changes.
- We describe 13 additional cases to expand our knowledge and to create awareness of this rarely reported condition.
- Fibrolipomatous hamartoma can be associated with macrodactyly; thus, it should be considered with the cutaneous syndromes associated with macrodactyly.

## RESULTS

### Clinical features

The clinical features are summarized in Table I. Patients' age at consultation ranged from 7 to 52 years (mean 29.5 years). Male and female patients appear to be equally affected, with a total of 7 female and 6 male patients. All patients presented with unilateral lesions on the thenar areas, fingers, or both. In 7 cases the lesions presented congenitally and had been slowly growing in size. In 6 cases the lesions presented sporadically and with no history of trauma before the onset of symptoms. All patients with congenital lesions noticed slight enlargement over the years together with mild symptoms such as paresthesia (numbness and tingling) and loss of strength. On physical examination the lesions consisted of a soft, firm, nonfluctuant, mobile, and minimally tender subcutaneous mass with a lobulated appearance located on the thenar and/or volar aspect of fingers. The affected fingers were greatly enlarged in size when compared with the contralateral digit (macrodactyly) (Fig 1, *A*). Six congenital cases presented with macrodactylia and 1 case had both macrodactylia and syndactylia (case 3) (Fig 1, *B*). It was usually the index and long finger that were enlarged in cases with macrodactyly (Fig 1, *C*). Patients had no positive family history of fibrolipomatous hamartomas, macrodactyly, or neurofibromatosis. Radiologic studies of the affected area revealed a soft-tissue mass without involvement of bone structures.

### Histologic features

All cases showed similar histopathology. On low magnification, all lesions showed interlacing collagen bundles, fibroblasts, mature adipocytes, and occasional capillaries mostly located in between nerve fascicles (Fig 2, *A*). The amount of fibrous to adipose tissue varied from case to case (Fig 2, *B*). In all cases, the nerve trunk was surrounded and infiltrated by fibroadipose tissue (Fig 2, *C*). The epineurium and perineurium was expanded by fibroadipose tissue that surrounded and separated the nerve bundles. Within the fibroadipose tissue a fibroblastic proliferation with perineurial fibrosis surrounded the nerve bundles. Several large mature nerves showed perineurial and endoneurial infiltration by connective tissue, small blood vessels, and adipose tissue. Multiple nerve fascicles with surrounding fibrovascular sheathlike structures were also seen. The nerve fascicles were well preserved; however, perineurial fibrosis was present at least focally in all of our cases (Fig 3, *A*). Also, in 7 cases there was neural degeneration and atrophy mostly caused by the prolonged compression of the nerves by the fibroadipose tissue. In addition, in 3 cases the nerves showed intraneural perineurioma-like features. In such cases, the nerves were expanded by the formation of small "onion bulbs" consisting of