Lipofibromatosis-like neural tumor: Case report of a unique infantile presentation

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A 14-month-old boy presented with a slow-growing, asymptomatic back plaque, which was biopsied and found to have S100 positivity, sparse CD34 staining, and no significant mitotic activity, nuclear pleomorphism, or necrosis; genetic workup found *LMNA-NTRK1* gene fusion, overall consistent with lipofibromatosis-like neural tumor (LPF-NT). LPF-NT is rare, with 14 cases previously reported, and our patient is the first report of this diagnosis in infancy. This case report and literature review includes comparison of similar diagnoses including lipofibromatosis, low-grade malignant peripheral nerve sheath tumor, infantile fibrosarcoma, and dermatofibrosarcoma protuberans and serves to aid detection of LPF-NT presenting in pediatric patients by highlighting similarities and differences that should prompt consideration. LPF-NT shows locally aggressive behavior only and should not be confused with conditions that have potential for distant spread. However, case reports of metastasizing *LMNA-NTRK1* tumors draw into question whether growths with this gene fusion exist on a spectrum of disease severity. Our patient was treated with wide local excision and has developed no complications or evidence of recurrence with 6 months of follow-up time. (J Am Acad Dermatol 2018;4:185-8.)

Key words: infantile mesenchymal tumor; lipofibromatosis-like neural tumor; pediatric skin tumor.

INTRODUCTION

Infantile mesenchymal tumors can range from benign to malignant, and proper diagnosis is crucial for patient management and counseling. Clinical appearance is not sufficient for diagnosis, and histopathology must be performed to determine tumor type. Lipofibromatosis-like neural tumor (LPF-NT) is a recently defined entity that commonly shows infiltrative growth and spindle cells arranged in streaming fascicles, which is similar to lipofibromatosis, but the tumor is distinguished by S100 protein reactivity and *NTRK1* gene rearrangements. Clinically, differential diagnoses other than lipofibromatosis include peripheral nerve sheath tumor, dermatofibrosarcoma protuberans, infantile fibrosarcoma, hamartoma, myofibroma, vascular plaque such as arteriovenous malformation, congenital nevus with proliferative nodules, and melanoma.

Abbreviations used:

LPF:	lipofibromatosis
LPF-NT:	lipofibromatosis-like neural tumor
FISH:	fluorescence in situ hybridization

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Conflicts of interest: None declared.

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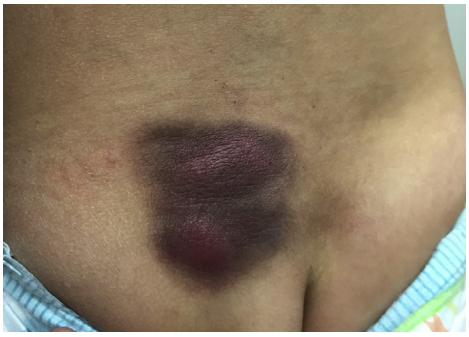

Fig 1. Lipofibromatosis-like neural tumor in a 14-month-old child, presenting as a hyperpigmented left lower back plaque.

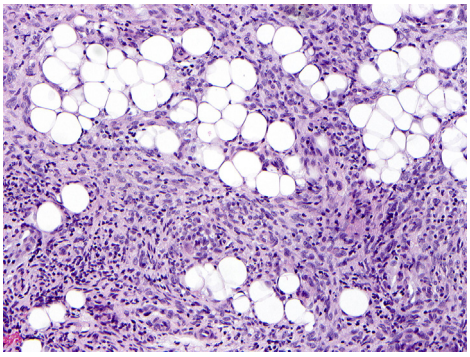

Fig 2. Lipofibromatosis-like neural tumor. Histopathologic sections of the skin biopsy specimen of this patient showed fascicles of spindled tumor cells infiltrating subcutaneous adipose tissue without significant cytomorphologic atypia or mitotic activity. (Hematoxylin-eosin stain; original magnification: ×20.)

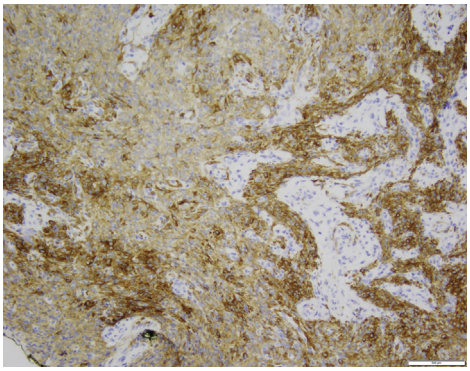

Fig 3. Lipofibromatosis-like neural tumor. NTRK1 immunohistochemistry of this patient's tumor shows positive staining in tumor cells (immunoreactivity indicated by brown chromogen). (Original magnification: ×10.)

REPORT OF A CASE

An otherwise healthy 14-month-old boy presented for evaluation of an asymptomatic truncal "birthmark" that slowly grew and changed color. He was born at 40.5 weeks via cesarean section after an uncomplicated pregnancy. Results of lower back ultrasound scan and radiography performed on the fifth day of life, because of to a small tuft of hair over the lower lumbar spine, were unremarkable. His parents reported the tumor to be present at birth, and his pediatrician documented a quarter-sized plaque at his 2-month visit. At age 9 months, the tumor measured 1.5 × 2 cm with central clearing. The plaque was not pruritic, painful, or friable.

With presentation to the dermatology department at age 14 months, physical examination found a 3- × 3.5-cm violaceous, hyperpigmented, atrophic plaque on his left lower back (Fig 1). It contained 2 prominent erythematous firm nodules, the larger nodule measuring 1.5 cm. Magnetic resonance imaging found a well-defined 3.8- × 3.4- × 0.6-cm discoid mass involving the skin and subcutaneous tissue with predominant T2 hyperintensity, intermediate T1 signal, and a small internal fat signal component.

Histology found a deep dermal and subcutaneous spindled-cell neoplasm with fascicular growth and infiltration into the adipose tissue but no significant mitotic activity, nuclear pleomorphism, necrosis, or hemangiopericytoma-like vascular proliferation (Fig 2). Tumor cells displayed focal S100 protein reactivity and very focal to weak CD34 staining but were negative for desmin, smooth muscle actin, epithelial membrane antigen, and anaplastic lymphoma kinase. Cytoplasmic NTRK1 immunohistochemistry showed diffuse positive staining (Fig 3), and fluorescence in situ hybridization (FISH) studies with custom Bacterial Artificial Chromosomes (BAC) probes found *NTRK1* breakapart. Further fusion FISH assays showed *LMNA-NTRK1* fusion, whereas testing for *ETV6* or *EWSR1* gene rearrangements was negative. Overall findings were most consistent with a diagnosis of LPF-NT.

The tumor was excised with 1-cm margins. The patient had a temporary vacuum-assisted closure to allow for confirmation of clear margins by formalin-fixed and paraffin-embedded pathology evaluation, and the defect was repaired with bilateral V-Y advancement flaps. The patient has no complications or evidence of clinical recurrence with 8 months of follow-up. Three months after excision, magnetic resonance imaging showed no definitive evidence of residual or recurrent tumor; this finding serves as a postoperative imaging baseline.

DISCUSSION

Lipofibromatosis (LPF) tumors, first described in 2000 by Fetsch and colleagues,[1] are rare,

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