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Management of advanced plexiform neurofibromatosis of the foot presenting with skeletal deformation and intractable pain: An indication for proximal amputation

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ABSTRACT

Plexiform neurofibromas of the foot are rare and often present with significant pain, deformity, and functional impairment secondary to their locally invasive behavior. While treatment has traditionally focused on attempts at radical resection, a lack of consensus among surgeons has hindered the establishment of a well-defined algorithm to guide the management of these highly co-morbid peripheral nerve sheath tumors. We present the case of an advanced plexiform neurofibroma of the right foot in a 24-year-old male with neurofibromatosis type 1. The patient presented following accelerated tumor growth with extensive osseous erosion, intractable pain, and progressive ankle instability that limited his capacity to ambulate and wear shoes. A modified transtibial amputation with a vascularized fibular bone graft (Ertl procedure) was performed without complication. Following graduated rehabilitation, postoperatively, the patient regained functional independence and was able to ambulate without pain in a customized prosthesis after 3 months. Plexiform neurofibromas of the foot present a complex challenge for foot and ankle surgeons. On the basis of our experience and previously reported cases, we advocate for amputation over aggressive attempts at advanced limb salvage for patients with extensive skeletal destruction, joint instability, and/or intractable pain caused by tumor mass effect.

1. Introduction

Plexiform neurofibromas (PN) constitute a significant source of structural and functional morbidity in patients with neurofibromatosis type I (NF1) [1,2]. Classically described as complex, benign peripheral nerve sheath tumors (PNST), PNs affect 20–40% of individuals with NF1 and are characterized by their diffuse and invasive growth patterns as well as their propensity for malignant degeneration (10%) [1,3]. Unlike schwannomas, PNs are intimately associated with functional nerve fibers and fascicles, passing centrally within the tumor itself. As a result of their locally aggressive behavior, patients often present with significant pain, physical deformity, and progressive neurologic dysfunction secondary to tumor growth and compressive mass effect [3,4]. If left untreated, these sequelae may have significant implications with respect to a patient's overall survival, functional prognosis, and quality-of-life over time.

While PNs can affect nearly any structure in the body, involvement of the foot is exceedingly rare with only seven reported cases to date [4–10] (Table 1). Management of these tumors has traditionally centered on radical resection, which is frequently complicated by extensive growth and invasion of adjacent vital structures, including major peripheral nerves [1–3,11–13]. In patients who are genetically predisposed to tumor formation, an expectant sense of futility and enhanced tolerance to symptomatology often confounds this situation by contributing to a delayed presentation in seeking treatment and reluctance toward surgery that may further compromise function [14,15]. As a result, NF1 patients often present with advanced disease, precluding complete resection. Attempts at marginal excision commonly herald more aggressive local recurrences with the potential for skeletal involvement and progression toward malignancy [3,9].

In this scenario, the best treatment option is the one that offers a superior functional outcome while minimizing the morbidity

Abbreviations: MRIm, agnetic resonance imaging; NF1n, eurofibromatosis type 1; PNp, lexiform neurofibroma.

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Table 1
Summary of previously reported cases of plexiform neurofibromas involving the foot.

Patient no.	Author, year	Age (years), gender	Location	Tumor size (cm^2)	Structures involved	Previous debulking	Treatment	Recurrence	Follow-up
1	Minton, 1980 [5]	20, F	Hindfoot	10	Skin, PTN	Yes	Radical resection	No	12 months
2	Harris and Sorto, 1981 [6]	8, M	Hindfoot	N/A	Skin, PTN	No	Radical resection	No	5 months
3	Turra et al., 1986 [7]	8, F	Forefoot	3	MPN, phalanges	Yes	Semiconservative resection	No	24 months
4	Blitz et al., 2002 [8]	20, F	Dorsum	42[a]	DPN, DP, PB	No	Radical resection	No	12 months
5	Pu and Vasconez, 2004 [4]	17, M	Heel/ankle	528	TP, FHL, FDL, PTA	Yes	Staged radical resection and FTT	No	18 months
6	Bourne et al., 2009 [9]	15, M	Heel	N/A	Skin, pedal skeleton	Yes	Below-knee amputation	N/A	N/A
7	D'Orazi et al., 2014 [10]	33, F	Sole/ankle	9	MPN	Yes	Radical resection	No	12 months

DP, dorsalis pedis; DPN, deep peroneal nerve; FDL, flexor digitorum longus; FHL, flexor hallucis longus; MPN, medial plantar nerve; N/A, information not available; PB, peroneus brevis; PTA, posterior tibial artery; PTN, posterior tibial nerve; TP, tibialis posterior.
[a] Value equals the sum of the area of two distinct tumors from the same region.

of local recurrence, protracted recovery, and/or premature loss of function. We present the case of an advanced PN of the foot resulting in pain and progressive ankle instability and discuss management strategies, including the indications for amputation over advanced limb salvage.

2. Case report

A 24-year-old male with NF1 and a complicated history involving complex maxillofacial reconstruction, following radical resection of a large PN of the face, presented with a progressively enlarging soft tissue mass along the medial plantar surface of his right foot. The mass had been gradually enlarging since childhood; however, an acute acceleration in the growth rate over the past 18 months had resulted in progressively worsening pain and varus hind-foot deformity, which had interfered with the patient's ability to ambulate and wear shoes. No prior attempts at resection or tumor debulking were undertaken. At the time of presentation, the patient was largely non-ambulatory secondary to intractable pain and progressive ankle instability.

On physical examination, a large 25 cm × 15 cm amorphous mass was noted along the instep and medial half of the right foot, extending from the posterior heel to the level of the metatarsophalangeal joints (Fig. 1). The skin was supple and soft without discoloration. A significant varus deformity of the hind foot was appreciated at rest with the majority of the patient's weight being supported by the lateral malleolus on attempted ambulation. Range of motion of the right ankle was restricted in all directions and limited largely by intense pain. The patient demonstrated hypersensibility to light touch and pinprick of the skin overlying the mass. The right foot was neurovascularly intact, with palpable dorsalis pedis, posterior tibial, and peroneal pulses. No pathologic reflexes were elicited. The remainder of the exam was significant for the presence of axillary and inguinal freckling, multiple subcutaneous neurofibromas, and several café-au-lait spots, consistent with a diagnosis of NF1.

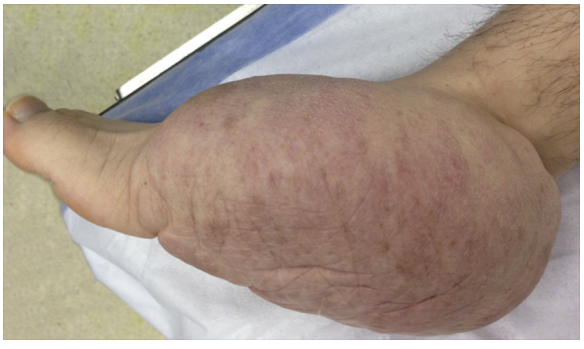

Fig. 1. Photograph of a large (25 cm × 15 cm) plexiform neurofibroma involving the medial surface of the right foot and posterior heel.

Preoperative plain-films showed extensive osseous erosion with distortion of the mid-foot and hind-foot architecture (Fig. 2A and B). Magnetic resonance imaging (MRI) demonstrated a large hypointense soft tissue mass encompassing the medial half of the foot and posterior heel on T1-weghted images and increased signal intensity on T2-weighted images (Fig. 2C and D). The mass appeared to infiltrate surrounding soft-tissue structures, including involvement of the posterior tibial vessels and tibial nerve. Extensive bony erosion of the mid-foot and hind-foot was appreciated. Findings on physical exam and imaging were consistent with a diagnosis of a large plexiform neurofibroma of the foot.

Given the presence of intractable pain and instability, along with the patient's desire to avoid a more protracted recovery with a high risk of recurrence or malignant transformation, the patient elected to proceed with a below-knee amputation. A modified Ertl procedure was performed, utilizing a vascularized fibular bone graft with fine wire fixation, as described previously [16]. A tibial osteotomy was performed approximately 15 cm distal to the tibial tuberosity, leaving a long posterior myocutaneous flap intact. The first fibular osteotomy was made distal to the tibial cut at a distance equivalent to the width between the lateral and medial cortices of the tibia and fibula, respectively. A second, proximal fibular bone cut was performed at the level of the previous tibial osteotomy. Great care was taken to preserve the peroneal artery attachments to the vascularized fibular bone bridge, which was fixed between the distal tibia and fibula using 24-gauge stainless steel sternal wires (Fig. 3A). Preservation of the lateral compartment musculature during the dissection allowed for additional soft-tissue coverage of the distal tibia and fibula (Fig. 3B). The posterior myocutaneous flap was designed such that the anterior soleus fascia was sutured to the anterior cortex of the tibia, and the distal Achilles tendon was apposed to the anterior crural fasica using multiple polydioxanone sutures prior to skin closure. Fixation of the bone strut was confirmed on postoperative X-rays (Fig. 4). The final pathology report confirmed the diagnosis of plexiform neurofibroma with no morphologic evidence to suggest malignant transformation.

Postoperatively, the patient was kept in knee-immobilizer for 4 weeks to prevent flexion contracture at the knee. A graduated rehabilitation program, consisting of non-weight bearing range of motion training and proximal limb strengthening was begun immediately. The amputation site healed uneventfully, and the