

Definitive Surgical Treatment of Lower Extremity Periungual Fibromas in a Patient with Tuberous Sclerosis

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STATEMENT OF PURPOSE

This case report highlights one adult patient with undiagnosed tuberous sclerosis whose manifestation of the disorder was limited to the lower extremities. Proper course of actions were taken clinically to make the correct diagnosis and plan for a definitive surgical treatment.

LITERATURE REVIEW

Tuberous sclerosis complex (TSC) is a rare, autosomal dominant, neurocutaneous disorder characterized by unexpected growths of normal tissue. The incidence of this genetic disease is estimated at 1 in 6000 live births. This results in the formation of benign tumors, usually affecting the brain, heart, kidney and lungs. The disorder may also display cutaneous manifestations, including benign periungual and subungual fibromas, hypopigmented skin macules, and facial angiofibromas. The pathophysiology of this condition involves mutation in TSC1 or TSC2 gene which provide instructions for making the proteins hamartin and tuberin, respectively. Within cells, these two proteins work together to help regulate cell growth and size^{1,3}.

While many patients exhibit symptoms at birth or early childhood, others may mask the disorder until adulthood. Early diagnosis is often triggered by central nervous system symptomatology, more specifically the formation of cortical tubers, or groups of malformed, abnormal cellular tissue within the brain. These malformations often result in seizure activity, leading to further workup and diagnosis of the disease. Others may develop angioliomas which consist of fat and muscles and these are usually a benign lesions that are most commonly seen on kidneys². While periungual fibromas are reported in patients with tuberous sclerosis, it is not as commonly seen. To our knowledge, no treatment protocols have been defined for proper treatment of excess soft tissue overgrowth due to the condition. Histologic analysis and tissue biopsy is recommended to rule out malignancy. Common histologic findings associated with cutaneous growths include abundant blood vessels, vertically oriented dense collagen fibers and fibrosis^{4,5}.

CASE STUDY

This case report examines a 72 year old male patient who presented to the office with periungual fibromas spanning the nail beds of all ten digits. Patient reported a longstanding history of these growths and denied any prior treatment or intervention. Upon clinical examination, the patient exhibited bullous exuberant hypertrophic skin lesions to the medial and lateral nail beds of all digits of both feet. Lesions were non-pigmented and appeared to extend beyond skin boundaries. Initial management involved performance of an in office biopsy of a small tissue sample of the left fifth digit. The pathology report generated a diagnosis of acquired digital fibrokeratoma within the volar skin of the fifth digit, consisting of thickened vertically oriented collagen bundles (Fig. 3). Upon relaying the diagnosis to the patient, the patient confirmed a multi-generation familial history of tuberous sclerosis. He denied ever having any prior workup to determine if he himself had the disease or carried the gene for TSC.



Figure 1: Clinical presentation, left foot



Figure 2: Clinical presentation, right foot

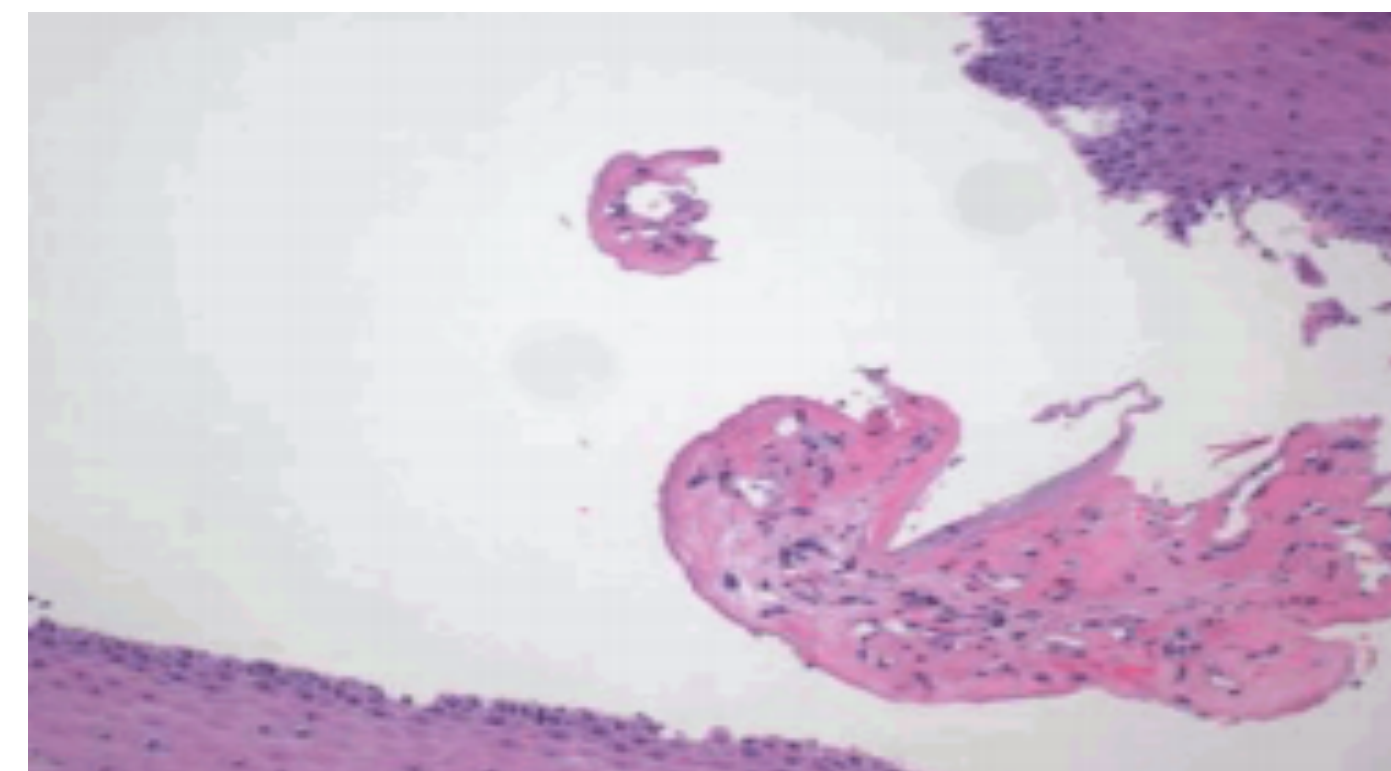


Figure 3: Hematoxylin-eosin stain showing vertically oriented collagen bundles with fibrosis

CASE STUDY (continued)

Due to the aggressive, widespread nature of the fibromas, the decision was made to definitively address these lesions with distal Symes amputations of the lesser digits, as well as soft tissue excision with nail bed cauterization to bilateral halluces. No recurrence was noted in the 21 month follow up period.

Upon surgical intervention, incisions were made overlying the dorsal interphalangeal joints of the lesser digits. Next, the distal phalanges of the lesser four digits of the left foot were resected and passed from the operative field. The entirety of the dorsal periungual skin, inclusive of the periungual fibromas, was resected, leaving the plantar glabrous skin intact for flap closure. Next, the lesser digits were flushed with sterile saline and the plantar skin was flapped dorsally and reapproximated to the healthy, dorsal skin, proximal to the site of the fibromas. To allow for adequate healing and patient function, the surgeon waited one month before performing the identical procedure on the lesser digits of the right foot. It is of note that the procedure performed to bilateral halluces strictly involved superficial debridement of periungual fibromas without osseous resection. This decision was made prior to surgical intervention, due to the essential function of the great toe in active propulsion and gait.



Figure 4: Intra-operative image. right foot

RESULTS

This case report demonstrates a 72 year old male patient with bilateral lower extremity periungual fibromas, consistent with tuberous sclerosis, successfully treated with digital distal Symes amputations of the lesser digits of bilateral feet. Following surgical intervention of bilateral feet, with a one month interval between surgeries, the patient healed uneventfully with no complications noted. Additionally, the patient was followed for a total of 22 months and no recurrence of fibrokeratomas was noted.



Figure 5: Post-operative image, left foot



Figure 6: Post-operative image, right foot

ANALYSIS AND DISCUSSION

Tuberous sclerosis, although rare, may present challenges to a patient's quality of life, even without causing organ-specific symptomatology. In the case of patients with aggressive skin overgrowths involving multiple digits, one should consider digital amputations to prevent recurrence and provide functional relief. It is also important to note that the surgeon used electrocautery at the base of the soft tissue prior to surgical closure. This may be an important factor which may prevent any recurrence or overgrowth of the lesions.

Of note, patients with tuberous sclerosis are prone to renal overgrowths such as angiomyolipomas, which can be fatal when they become enlarged⁶. Making the diagnosis can allow the podiatric provider to appropriately treat the cutaneous manifestations, as well as properly refer the patient for additional systemic workup⁷.

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