Cutaneous Myofibromatosis Masquerading As A Chronic Non Healing Leg Ulcer
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INTRODUCTION
Myofibromatosis, congenital generalized fibromatosis or infantile fibromatosis (IM) is a mesenchymal disorder of early childhood where in there are multiple fibrous nodules in soft tissues and internal organs. It was thought to be a rare condition that was frequently fatal. Because the lesions may not be easily discernible and do spontaneously resolve, this condition is under diagnosed and under reported. Kauffmann et al grouped these patients into two categories: (1) a multiple form, with lesions restricted to skin, subcutaneous tissue, skeletal muscle and bone with a good prognosis and (2) a generalized form, with visceral lesions and poor prognosis. Following recognition of the myofibroblastic nature of the constituent cells, Chung et al reported 61 cases of this entity and named it IM. Our patient presented with a non healing ulcer on the left lower limb and posed a real problem in the diagnosis initially as we never thought of a possibility of myofibromatosis and also as it is an extremely rare entity encountered in day to day surgical practice as a cause of non healing leg ulcers.

CASE HISTORY
An eight-year-old boy presented with multiple nodules in the lower aspect of the left lower limb of six months duration, the largest one being ulcerated since two months (Figure 1). Though the ulcer was non healing it had neither spread nor increased in size. Two new lesions had appeared one week back. There was no history of trauma, varicose veins or arterial insufficiency. The family history was not contributory. A general examination was normal. Local examination revealed multiple subcutaneous nodules of various sizes, which were mobile and non tender. There was a serous discharge from the ulcerated nodule, which measured 2x1 cm. There were no similar lesions in the rest of the limb, opposite limb or any where else on the body. There was no inguinal lymphadenopathy, varicosities and all the peripheral pulses were felt. Systemic review was unremarkable.

Figure 1
Figure 1: Photograph showing multiple nodules on the left lower limb

A clinical diagnosis of cutaneous tuberculosis was made as the lesions had a chronic, non healing course with appearance of few new lesions. Routine investigations [complete blood examination including an erythrocyte sedimentation rate (ESR), chest x-ray (CXR)] were normal. Mantoux test was negative. Culture of the ulcer discharge grew no organisms. An excision biopsy of a nodule was executed, which revealed proliferation of spindle cells with criss-crossing bundles showing thin spindly nuclei and eosinophilic cytoplasm and abundant collagen which was
reported to be myofibromatosis (Figure 2) and was reinforced by special stains (positive staining for smooth muscle actin). Further to exclude additional lesions, an ultrasound scan (USG) of abdomen and echocardiogram (ECHO) was done and were found to be normal.

**Figure 2**
Figure 2: Photomicrograph showing spindle cell proliferation in a whirling fascicular pattern (H & E, x20).

Patient was conservatively managed with bed rest, daily dressings and was discharged as the lesions showed satisfactory regression in a week's time. He was kept under close follow up and after eight months there has been a complete regression of the lesions and remains asymptomatic.

**DISCUSSION**

Myofibromatosis is a rare disorder and has been a known entity since 1954. Most of these tumors are readily identifiable by their clinical manifestations and histological appearance. Characterization of myofibromatosis includes the formation of firm to hard masses in skin, muscle, viscera, bone and subcutaneous tissue. The typical lesion is a single (less often, multiple), firm, subcutaneous dermal nodule. Cutaneous lesions are most common in the head and neck region. Chung et al coined the term IM in order to emphasize the microscopic resemblance of the lesion to smooth muscle tissue and the frequent occurrence of the disorder in both newborns and older infants.

Solitary disease is more common in males, whereas patients with multifocal disease, which more commonly affects females, may have few to hundreds of lesions. Such patients may also have myofibromas develop within the skeleton, myocardium, gastrointestinal tract, lungs or a combination of these. The most common extracutaneous location is bone and is involved in greater than 50% of cases of multifocal myofibromatosis. Extensive visceral involvement, especially of the pulmonary and gastrointestinal tract, heralds a poor prognosis. The tumors are usually rubbery in nature and vary from 0.5 to 3 cm in diameter. A single large tumor with multiple smaller lesions may be mistaken for a primary malignancy with metastases. Due to prominent tumor vascularity, skin lesions may resemble hemangiomas. When pulmonary involvement is present, a CXR may be suggestive of diffuse interstitial inflammation or bronchopneumonia.

The cause of this condition is unknown. Myofibromas are best regarded as developmental anomalies rather than true neoplasm. Although most reported cases are sporadic, several cases of familial disease have been reported. Bartlett et al first reported a family in whom autosomal recessive inheritance was suspected because of consanguinity between parents. Bracko et al has suggested autosomal dominant inheritance, which has been supported by Zand et al. Additionally, investigators have proposed that these lesions may be hamartomatous or might result from in utero stimulation from estrogenic hormones.

The characteristic histopathology of a myofibroma shows a dermal or subcutaneous infiltrate of spindle-shaped, fibroblast-like cells and fusiform, smooth-muscle like cells arranged in a whorled pattern. There is also abundant collagen within the surrounding matrix. Necrosis, hyalinization and calcification may be present.

Myofibromatosis must be differentiated from neurofibromatosis, fibrosarcoma, hemangiopericytoma and leiomyoma/leiomyosarcoma. The typical presentation and characteristic histological features serve to easily differentiate myofibromatosis from these other disorders.

Evaluation of patients with myofibromatosis should include a thorough family history, a complete physical examination (to search for additional lesions), CXR, USG of the abdomen, ECHO and tumor biopsy.

Death from myofibromatosis is only seen due to complications i.e., cardiopulmonary or gastrointestinal, in the multicentric form of disorder. Otherwise it is a benign, self limited disorder. The typical course is that of spontaneous regression, as in our patient. With advanced supportive care, patients with severe visceral disease may have favorable outcomes without treatment. However, for patients with affected vital functions and life-threatening disease surgical treatment is indicated. Chemotherapy or
interferon Alfa, with or without radiation, has also been used successfully for patients with unresectable tumors.

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