# Giant recurrent infantile myofibromatosis of the leg in a 3 year-old

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## ABSTRACT

Infantile myofibromatosis is a rare mesenchymal disorder of infancy and childhood characterised by the formation of tumours in the soft tissues, muscles, bones and viscera. Only 61 cases have been reported in the literature. This paper describes the management of a 3 year-old boy with extensive IMF of the right leg.


**Key words:** Infantile myofibromatosis, children, management

## INTRODUCTION

Infantile myofibromatosis (IMF) was first described in 1981 by Enzinger in a study of 61 cases of congenital and infantile myofibromatosis.[1] It is a rare mesenchymal disorder of infancy and childhood characterised by the formation of tumours in the soft tissues, muscle, bone and viscera, and usually presents before the age of 2 years.[2] The disease is limited to the soft tissues, muscle and bone. It has a good prognosis, and excision is curative; however, visceral involvement may be fatal.

Diagnosis of IMF is difficult because of the clinical heterogeneity and the histopathological appearance. The diagnosis relies on identification of two separate tissue components namely, fascicular myofibroblastic at the periphery and haemangiopericytome in the centre.

## CASE REPORT

A 3 year-old boy presented to us with a huge recurrent IMF of the right leg, which had been growing very rapidly of late. The mass had been present since birth, but was incompletely excised a year ago in another centre.

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Examination showed a marasmic boy in a very poor general condition. The mass appeared malignant clinically, and extended from the knee to the ankle [Figure 1].

Investigations done included a plain X-ray and computed tomography (CT) of the lesion, which showed infiltration of the muscles of right leg and erosion of the bones [Figure 2]; CT of the chest revealed no infiltrations or secondary deposits [Figure 3].

The boy was prepared and later had an above knee amputation because the bones of the right leg had been destroyed by the growth. Histopathology confirmed the diagnosis of IMF with a mitotic index of 3. The outcome was uneventful and satisfactory.

## DISCUSSION

Myofibromatosis is a rare mesenchymal disorder of infancy and childhood. Solitary lesions are more common and have a marked tendency to occur in boys, while the less common multicentric disorder predominantly affects girls.[1] Approximately 90% of cases present by two years of age, and its occurrence in older children is unusual. IMF has a familial tendency and carries a good prognosis when solitary but death



**Figure 1: Infantile myofibromatosis of the right leg**

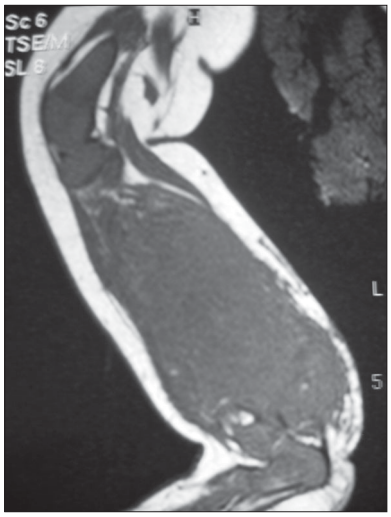



**Figure 2: Computed tomography of the right leg**

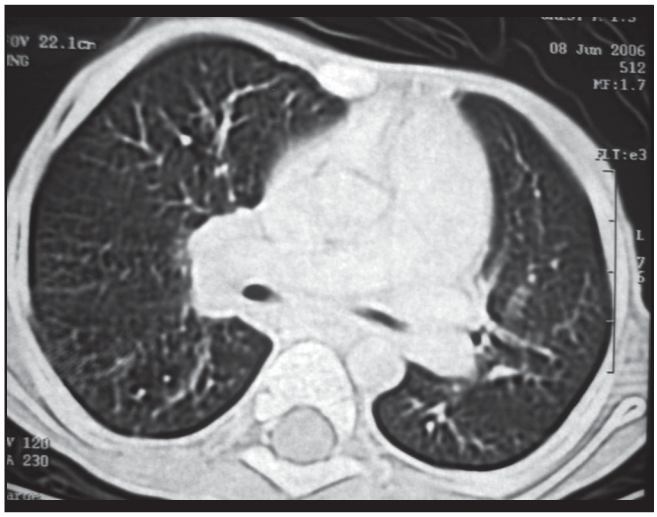



**Figure 3: Computed tomography of the chest**

frequently occurs in generalized IMF with visceral involvement.

Diagnosis of IMF is difficult because of the clinical heterogeneity, variable radiological (which often mimics an aggressive neoplasm) and histopathological appearance. The diagnosis relies on identification of two separate tissue components; fascicular myofibroblastic at the periphery and hemangiopericytome in the centre. Histologically, non-malignant tumours must be differentiated from malignant ones since their management and evolution is completely different.[3]

It is a potentially life-threatening entity and should be considered in the differential diagnosis of pediatric dermal and subcutaneous nodules, particularly when associated with a recent onset of head tilt. Close clinical follow-up is recommended in all cases of infantile myofibromatosis.[4] Arcangeli and Calista [5] reported congenital myofibromatosis in two siblings, from non-consanguineous parents; the large-sized subcutaneous myofibromas were surgically removed, and the smallest ones disappeared spontaneously over the course of 24 months.

Since spontaneous tumour regression is expected in cases of limited involvement, therapeutic abstention and patient observation are recommended. Surgery, chemotherapy or radiotherapy is reserved for patients with multiple visceral localisations because the later carry a bad prognosis with a potentially fatal outcome. The solitary form of IMF is most common and its treatment is complete excision with an excellent prognosis,[6,7] In extensive forms, conservative surgical excision is recommended because recurrence rates are low and there is a possibility of spontaneous regression.[2] An above knee amputation was performed in this report because the bones of the affected leg were already destroyed by the growth. Low dose chemotherapy is recommended in cases of generalised IMF, with life-threatening visceral and nonvisceral involvement.[8]

Recurrence is a potential complication in extremity cases presenting above the age of 5 years, parti-cularly if CT scan shows calcification and erosion of adjacent bone or if surgical resection is income-plete. Mitotic index of 5 or more per 10 high-power fields and the presence of areas of necrosis and inflammation within the tumour are bad prognostic findings associated with a high incidence of recurrence.[9,10)]

Infantile myofibromatosis should be considered in the differential diagnosis in any child who presents with either a solitary or multiple tumours, particularly those occurring in the first 2 years of life.

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