CASE REPORT

Infantile Myofibromatosis

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Summary

Infantile myofibromatosis (IMF) is a rare tumour with a wide spectrum of disease activity ranging from a solitary cutaneous nodule through to a multicentric form with widespread visceral involvement. It is characterised by its unique ability to spontaneously regress and has a typical histological appearance of actin-positive fibroblasts arranged in whorls or fascicles and vessels in a pericytomatous pattern. A male infant with multiple lesions involving the subcutaneous tissue and bone from birth is described and followed-up for two years. Treatment of IMF is dependent on the location of the tumour/s with surgery or chemotherapy reserved for rapidly progressive or symptomatic disease. However, due to the low rate of recurrence and the possibility of spontaneous tumoral regression, therapeutic abstention, as practised in our patient, is justified.

Key Words: Infantile myofibromatosis, Congenital mesenchymal tumours

Introduction

Infantile myofibromatosis (IMF) is an uncommon, usually cutaneous, condition in which there is a benign proliferation of myofibroblasts. Solitary and multicentric nodular forms with, and without, visceral involvement have been described. Infantile and adult sub-types have been reported, each having distinct clinicopathological features. Presentation in the head and neck is common. It is frequently misdiagnosed because of its peculiar histological features and must be differentiated from a malignancy to avoid unnecessary administration of chemotherapy or radical surgery.

Case Report

Our patient was first noticed to have firm lumps, measuring approximately 2 x 2cm each, at his right elbow and lower back at birth. As there was no overlying skin abnormality and these lumps did not appear to be tender, the parents did not seek further medical attention. The child was otherwise completely well and thriving. Over the following 3 months, these lumps progressively decreased in size and finally disappeared, each leaving a small, fibrous scar.

At the age of 5 months, two similar lumps appeared on his head. One measuring 4 x 4cm was located on his forehead, while another measuring 2 x 2cm was located at the occiput. The parents sought medical attention at their local hospital where fine needle aspiration biopsy was performed. Spindle-shaped cells were seen and a diagnosis of embryonal rhabdomyosarcoma was made. He was then referred to the University of Malaya Medical Centre (UMMC) for further management.

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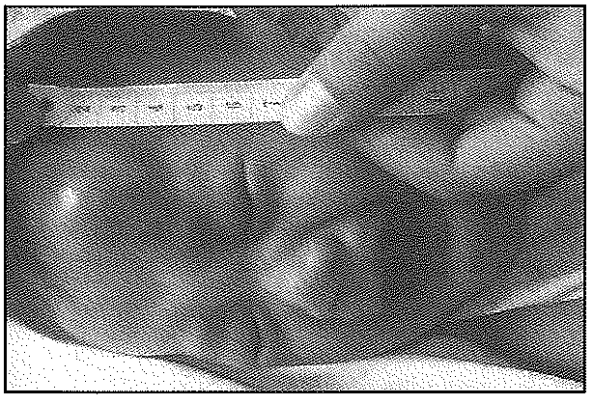

Fig. 1: Infantile Myofibromatosis.

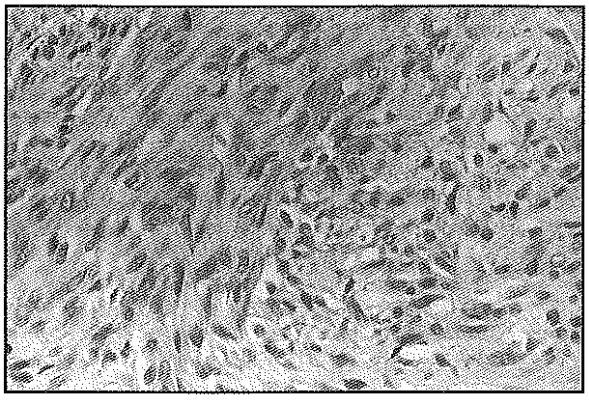

Fig. 2: Infantile Myofibromatosis.

At UMMC, he was found to have the two firm, subcutaneous lumps which were not adherent to the overlying skin (Fig 1). He was thriving (all anthropometric measurements were between the 75th to 90th percentiles for age) and systemic review was normal. Investigations done were as follows: Haemoglobin 121g/L, platelets 319 x 10^9/L and WBC 9.5 x 10^9/L. Radiological skeletal survey showed radiolucent lesions at the proximal metaphyses of his left humerus and frontal bones.

A wedge biopsy was done on the forehead lump. Histopathological examination showed the tumour was composed of spindle-shaped cells having elongated oval vesicular nuclei with prominent nucleoli and indistinct cell borders arranged in whorls and in some places showing a hemangio-pericytomatous pattern [Fig 2]. The mitotic activity was mildly increased (5/10hpf). Special stains showed that the tumour cells expressed strong cytoplasmic immunopositivity for actin but were weakly positive for myoglobin and negative for desmin and S-100 protein. These findings, supported by the clinical history of spontaneous regression of earlier swellings, were consistent with a diagnosis of IMF. He was then treated conservatively with close follow-up.

Over the following 8 months the forehead lump grew bigger before plateauing at 5 x 5cm. A new small lump developed over the upper lip, 1 x 1cm while the lump at the occiput remained the same size. However, he remained asymptomatic and well. At two years' follow-up, the forehead lump had regressed to a flat plaque measuring 0.5 x 0.5cm while both the lumps at the occiput and at the upper lip had disappeared. The child was well and thriving. Repeat radiological examination of the affected bones showed complete resolution of all lesions.

Discussion

IMF is a rare mesenchymal disorder of childhood. Approximately 250 cases have been reported in the literature since this condition was first described by Stout in 1954 but it is probably under-estimated due to its fairly inocuous nature. These tumours can occur as a solitary lump or be multi-centric. The commonest cinical presentation of IMF is the presence of firm, discrete subcutaneous nodules with the majority occuring in the head and neck region. In the multi-centric form, there can be numerous tumours, ranging from two to 100, involving virtually any organ in the body including the central nervous system.

The aetiology of IMF is unknown. It has been postulated that the foetus is affected in utero by stimulation by maternal oestrogens[2]. There have also been reports on IMF occurring in siblings or successive generations in families suggesting that it is a heritable condition[3].

The diagnosis is suggested by a history of spontaneous regression of the tumour(s), which is often present at birth, and confirmed on close histological examination and immuno-histochemical features. The combination of fibrous and angiomatous patterns in myofibromatosis may be confusing when examining a small biopsy, as with specimens obtained via fine needle aspiration. This may, in turn, lead to an erroneous diagnosis of malignancy as was initially thought of in our patient.

IMF confined to the skin, soft tissue and bone are associated with a very good prognosis due to their propensity for spontaneous regression[1]. However, there is no common time frame whereby regression is expected to occur and these tumours may even initially enlarge before becoming smaller[2,3]. This behaviour was observed in our patient. Occasionally, these tumours may even recur 15 to 25 years later. Nevertheless, when no life-endangering pressure on vital organs exists, these tumours should be dealt with conservatively; avoiding radical surgery or chemotherapy[2]. However, when there is involvement of the viscera or compression of vital organs, the prognosis is poor. In a review of 60 patients with multi-centric IMF (total 170 patients) by Wisewell, a mortality rate of 75% was reported. Deaths were related to complications of visceral involvement, usually cardiopulmonary or gastrointestinal[3]. In these cases, surgery with and without chemotherapy (ifosfamide, actinomycin and vincristine) or radiotherapy have been utilised. However, the non-surgical methods have been mainly anecdotal and success rates have been difficult to evaluate accurately owing to the small numbers involved.

In conclusion, IMF is a rare tumour with distinct clinico-pathological features. Accurate diagnosis relies on adequate tissue for histological examination and conservative management in cases confined to the skin and subcutaneous tissue appears justified.

References

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