Case report 684

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Radiological studies

Fig. 1. Radiograph obtained 1 year prior to current presentation. Note a soft-tissue mass at the proximal and medial aspect of the left thigh (arrows), with blurring of the adjacent femoral cortex.

Fig. 2. Radiograph from current presentation. The soft-tissue mass has increased in size and shows speckled calcification. Erosion of the femoral cortex is more pronounced, but no periosteal reaction is seen.

Clinical information

This otherwise healthy, 4-year-old boy presented with a history of a limp for 12 months. Clinical examination revealed a firm mass in the left groin, occupying Scarpa's triangle. The mass was observed on roentgenograms of the pelvis to be a soft-tissue mass with scattered densities and erosion of the proximal end of the femur (Fig. 2). A similar but less distinct soft-tissue abnormality with equivocal early osseous change was noted on radiographs obtained elsewhere 1 year earlier and made available to us (Fig. 1). The greatest diameter of the soft-tissue mass on the radiographs was thought to have increased when compared with the initial film from 5 to 6.5 cm. Bone scan showed slightly increased isotope uptake in the region of the proximal end of the left femur. Chest films and routine laboratory investigations were normal. At open biopsy the tumor was seen to be made of gray, soft, and very vascular tissue, with a network of large, distended vessels on its surface. The tumor bled freely from the biopsy site. Its fleshy macroscopic appearance reinforced the clinical and radiologic suspicion of a sarcoma.

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Diagnosis: Infantile myofibromatosis

Frozen section biopsy suggested infantile myofibromatosis. In view of the possibility of spontaneous regression associated with the frozen section diagnosis and the considerable technical difficulty presented by the vascularity and the size of the lesion, a decision was made against local excision. The diagnosis was subsequently confirmed on paraffin sections. The postoperative course was uneventful. An abdominal CT revealed no visceral lesion. When reviewed 6 months later the lesion appeared to be unchanged, although the preoperative clinical and radiological impression favored a primary soft-tissue tumor such as aggressive fibromatosis, fibrosarcoma, malignant fibrous histiocytoma, rhabdomyosarcoma, or synovial sarcoma.

Histology

The biopsy (Fig. 3) showed a moderately cellular tumor of uniform, cytologically bland spindle cells arranged in small fascicles or in a pericytic pattern in relation to a rich network of thin-walled, sinusoidal vascular channels. Many of the spindle cells, especially those in the fascicles, resembled smooth muscle cells. Mitoses were sparse. The typical zoning of various patterns is not seen, probably due to the small size of the biopsy. Electron microscopy study revealed the spindle cells to be a mixture of fibroblasts and myofibroblasts.

Discussion

Infantile myofibromatosis, formerly known as congenital generalized fibromatosis, generalized hamartomatosis, multiple congenital mesenchymal hamartomas, and multiple vascular leiomyomas [5], is a mesenchymal tumor in which most of the proliferating cells have the ultrastructural characteristics of myofibroblasts. The condition occurs in two clinical forms – multicentric and solitary.

With very rare exceptions the multicentric form is confined to neonates or young infants and can be further divided into somatic and somatovisceral subtypes. The somatic form involving the subcutis and musculature with or without skeletal lesions has a good prognosis with eventual regression of the lesions; involvement of the viscera is usually attended by a fatal outcome. Very unusually, the central nervous system is involved [1, 3]. Although 2–3 times less common than the solitary form [5], the multicentric variety is much more familiar to clinicians, often with a spectacular radiographic appearance in the larger number of case reports. Very occasionally this form is associated with multiple congenital malformations [8]. Familial occurrence is not unknown, and an autosomal dominant inheritance has been suggested [6].

The solitary form is also predominantly a disease of the 1st year of life but does occur in early childhood and sporadically in adolescence, young adulthood [5], and even old age [4]. Nearly all the lesions are in the peripheral soft tissue or bone, but rare, solitary, visceral lesions have been reported [10, 11].

Chung and Enzinger [3] reviewed 45 patients with solitary and 16 with multicentric lesions. Some 88% of these cases were diagnosed in the 1st year of life. The head, neck, and trunk were the most commonly affected sites. Only 5 of the 45 patients with solitary lesions had bone involvement, all in the cranium. The localized form of infantile myofibromatosis lacks distinctive clinical or radiographic features and is virtually always diagnosed only on histological study. The relatively advanced age of our patient and the location of his solitary lesion in an extremity, with erosion of a long bone, are unusual presenting features for this condition.

Because of the adjacent bony change, all our preoperative clinical and radiological differential diagnoses were those of either aggressive or malignant conditions, mainly of soft tissue. These included aggressive (desmoid) fibromatosis, fibrosarcoma, synovial sarcoma, and rhabdomyosarcoma.

The juxtaosseous form of aggressive fibromatosis is most common in the 1st decade of life [2], with girls more commonly affected than boys. In 50% of cases the lesion is localized

Fig. 3. Photomicrograph of biopsy. Uniform, bland spindle cells are noted in a partly collagenized background, disposed in small fascicles or around thin-walled, sinusoidal, vascular channels in a pericytic fashion (H&E × 36)