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Radiological studies

Fig. 1. A PA roentgenogram of the right leg at the age of 8 weeks demonstrates two well-defined ovoid lesions in the upper diametaphysis of the tibia and one larger lesion in the lower metaphysis of the same bone. Each lesion has a thin sclerotic margin with a central focus of mineralization (calcium or bone). B Three month later one of the two lytic lesions in the upper diametaphysis on the lateral aspects has virtually disappeared. The more medially placed of the two lesions in the upper diametaphysis is now smaller than on the study 3 weeks previously and less apparent, indicating healing. The lower metaphyseal lesion is now considerably less lucent, again suggesting healing.

Clinical information

This 8-week-old male infant was brought to the hospital for treatment of an upper respiratory infection. The physical examination was normal except for the presence of three firm, nontender, soft tissue nodules, one each in the right upper and lower limbs, and the third in the anterior abdominal wall. These findings led to a radiological skeletal survey which showed several well-circumscribed, ovoid, longitudinally-oriented radiolucent lesions in the diametaphyseal (and metaphyseal) regions of the left femur (not illustrated) and right tibia — the only bones affected (Fig. 1A). These lesions were not associated with the soft tissue nodules. Each bony lesion was characterized by a thin, sclerotic margin, a central focus of mineralization, absence of periosteal reaction and no cortical disruption or involvement of soft tissues. A radionuclide bone scan was negative. Several biopsies were performed. Three months later the lesions of the lower limbs had undergone evidence of considerable healing (Fig. 1B) and by the age of eight months all the skeletal soft tissue abnormalities had disappeared.


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Diagnosis: Congenital multiple fibromatosis

The tissue obtained at biopsy originated not from the skeletal lesions but from the subcutaneous soft tissue nodules. Histological studies (Figs. 2A and B) showed both mature and immature proliferation of fibrous connective tissue, interspersed between muscle bundles in some areas. Small foci within the lesions were composed of a hyalinized, chondroid-like matrix, surrounded by circumferentially-arranged, plump fibroblasts. Necrotic foci were present as were numerous vascular spaces running between bundles of fibrous connective tissue. Only rare mitotic figures and minimal atypia, consistent with regenerating cells, were present. No evidence were noted to support a diagnosis of malignancy. The histological changes were considered typical of congenital fibromatosis; electronmicroscopy demonstrated ultrastructural features consonant with that diagnosis.

The combination of rapid, spontaneous repair of well-circumscribed skeletal lesions and a negative radionuclide bone scan suggests the diagnosis. The radiological differential diagnosis includes cystic hemangiomatosis or lymphangiomatosis (lesions usually larger), lipomatosis (often calcified), metastases from neuroblastoma (more aggressive), neurofibromatosis (discrete skeletal lesions are rare), and Letterer-Siewe disease (more aggressive and often associated with skin lesions).

Other reported cases have demonstrated the bland necrosis occurring in association with stippled calcification. The extent of vascularity is variable, and some lesions have featured a plethora of thin-walled blood vessels, vaguely suggesting hemangiopericytoma. The lytic lesions within bone exhibit a histological pattern similar to that obtained from the lesions in the subcutaneous tissues, skeletal muscle and viscera in cases of the lethal form of the disorder.

Congenital fibromatoses encompass at least two, and possibly three different forms. Congenital generalized fibromatosis was the term given by Stout to a rapidly fatal disease, characterized by

Pathological studies

Fig. 2A A photomicrograph of the biopsy specimen obtained from a soft tissue nodule (H and E stain ×100) demonstrates the presence of hypocellular nodules of fibroblasts and more intensely cellular interspersed stroma. Thin-walled blood vessels are moderately abundant