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Radiological studies

Fig. 1A–C. Multiple, eccentric, sharply-defined lucent lesions are noted to involve the metaphyses of the femora, the proximal metaphysis of the right humerus and the proximal metaphysis of the left radius. Minimal sclerosis around these lesions is present. No soft tissue abnormalities are detected.

Clinical information

This 28-day-old female infant was noted to have decreased motion of the right lower extremity. The infant was the product of an uncomplicated full-term pregnancy with normal spontaneous delivery. The infant's Apgar score was low and the baby was treated in the intensive care unit for two days for respiratory depression and hypoglycemia, presumably due to maternal Demerol administration just prior to delivery. The infant recovered uneventfully and continued to thrive. Physical examination showed a vigorous responsive neonate with no abnormalities of the skin or extremities. The right hip was held in approximately 30° flexion, but could be extended passively to a neutral position. No soft tissue masses were noted and no evidence of subluxation of the hip joint was detected.

Plain films of the pelvis showed multiple, well-defined, radiolucent lesions involving the metaphyses of the femora (Fig. 1A). A skeletal survey was then obtained which demonstrated similar lesions involving the right humerus and left radius (Fig. 1B, C). Anteroposterior and lateral roentgenograms of the chest were normal. Abdominal ultrasound studies and laboratory studies were within normal limits.

A biopsy was performed.

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Diagnosis: Infantile myofibromatosis

The differential diagnosis included histiocytosis X (Letterer-Siwe disease), angiomatosis, hematogenous osteomyelitis, metastatic neuroblastoma, neurofibromatosis, polyostotic fibrous dysplasia, multiple non-ossifying fibromas, enchondromatosis and diffuse lipomatosis.

The biopsy of the specimen histologically showed spindle cells arranged in a whorled, collagenized background. Although most cells were reminiscent of fibroblasts, cells with cigar-shaped nuclei, more typical of smooth muscle, were noted.

Discussion

This disorder was first described by Stout in 1954 as “congenital generalized fibromatosis” in which multiple fibrous tumours of the subcutaneous tissues, muscle and viscera occur [1].

Pathologically, the lesion consists of fascicles of plump spindle cells, which are intermediate in histological properties and in staining features between fibroblasts and smooth muscle cells (Fig. 2). Ultrastructurally, the cells also have the characteristics of smooth muscle cells and of fibroblasts. These features are characteristic for fibromatosis in general, and, in this case, of infantile myofibromatosis. This entity is believed to originate in utero as the lesions become manifest at the time of birth or shortly thereafter. The etiology of the disorder remains obscure. Maternal infection, over-production of estrogen and trauma have been implicated [2-4].

Although the lesions initially are apparently neoplastic, their spontaneous resolution suggests that the process is essentially self-limited and benign, unless the initial lesions have involved the viscera to such an extent that malfunction incompatible with survival occurs. Morettin et al. in 1972 [2] classified this disorder into two forms: 1) a somatic form, with involvement limited to the subcutaneous and muscular tissue, with or without skeletal lesions; and 2) a combined somatico-visceral form which, in addition, has a more or less disseminated type of involvement. This classification helps to define the prognosis. The somatic form has a benign course leading to complete resolution both of the cutaneous and skeletal lesions. The combined somatico-visceral form has a poor prognosis with death in the first months of life. This form actually behaves in a manner similar to cystic angiomatosis with visceral involvement. This case belongs to the somatic form since the disorder is only localized to the bone. Therefore, it can be expected to be self-limited with an excellent prognosis. There are reports in the literature in which fibromatosis involved only the skeleton as in the case presented [4-8]. Spontaneous regression of fibromatosis usually takes from a few months to a few years [4, 9-14], and is well-documented in the literature.

The somatic muscular and subcutaneous lesions are widely disseminated, but it appears that a predilection for the proximal portions of the limbs exists. The subcutaneous nodules arise from or invade the underlying skeletal muscles or fascia. The somatic involvement may be limited to a single