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INFANTILE MYOFIBROMATOSIS: A CASE WITH UNUSUAL FEATURES AND REVIEW OF THE LITERATURE*

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SUMMARY: Kotiloğlu E, Göğüş S, Ruacan Ş, Akyüz C, Büyükpamukçu M, Sarıalioğlu F. (Pathology and Oncology Units, Department of Pediatrics, Hacettepe University Faculty of Medicine, Ankara, Turkey). Infantile myofibromatosis: a case with unusual features and review of the literature. Turk J Pediatr 1996; 38: 527-532.

A three-month-old boy was admitted for a slowly progressing nodule in his right chin, first recognized at birth and recently accompanied by facial paralysis. It was found to be a soft tissue mass arising in subcutaneous tissue and extending deep into the temporal muscle, causing temporal bone erosion without infiltrating the dura. Initially interpreted as mesenchymal chondrosarcoma, combined chemotherapy (PULSE-VAC) was given. Eighteen months later an additional nodule developed in the paraspinal skin. Evaluation of both lesions showed vimentin and α-smooth-muscle-action positivity. As the final diagnosis was multicentric infantile myofibromatosis, the child was followed up without therapy. Thirty months later, multiple osteolytic lesions appeared on the skull and long bones of the extremities. Some lesions remained stable and some regressed during two years of follow-up. *Key words: myofibromatosis, mesenchymal hamartomas, immunohistochemistry, childhood tumors.*

Childhood mesenchymal tumors of fibroblastic and/or myofibroblastic deviation are an adverse group of tumors with pathological features occasionally causing uncertainty about clinical behavior, treatment and prognosis. Infantile myofibromatosis (IM) is the most frequent tumor type among these, but it is not widely known and has been frequently misdiagnosed[1,2]. Here we report a case with an unusual course and review the relevant literature to familiarize both physicians and pathologists with the unique features of this tumor.

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Case Report

A three-month-old male was admitted in April 1990 for evaluation of a mass in his right chin, first noticed at birth. Its growth had been slow and progressive. One month prior to admission, signs of facial paralysis were noticed by his parents while he was crying. He was born at term as the first child of healthy parents with no consaguinity. There was no history of similar lesions in the family.

On physical examination, the tumor was palpated as an immobile, non-tender and firm nodule about two centimeters in diameter. A computerized tomography (CT) scan revealed temporal bone erosion without involvement of the temporal lobe. Other physical and laboratory findings were normal.

The tumor was totally excised along with the temporal muscle that it invaded. Both the fascia and the dura were intact. Microscopically the tumor was composed of spindle-shaped cells arranged interlacing fascicles at the periphery with myxoid and chondroid areas, especially in the center (Fig. 1). Mitotic figures were frequent (2-3/high-power field). The initial diagnosis was mesenchymal chondrosarcoma. It was accepted as stage II disease and combined chemotherapy (PULSE-VAC) was initiated[3]. Because of the patient's age, low doses were administered (Vincristin: 1 mg/m^2, Actinomycin-D: 10 μ/kg, Cyclophosphamide: 10 mg/kg); radiotherapy was not given. This protocol was carried out for one year with no problem. Several CT scans and pulmonary x-rays taken during this period were normal.

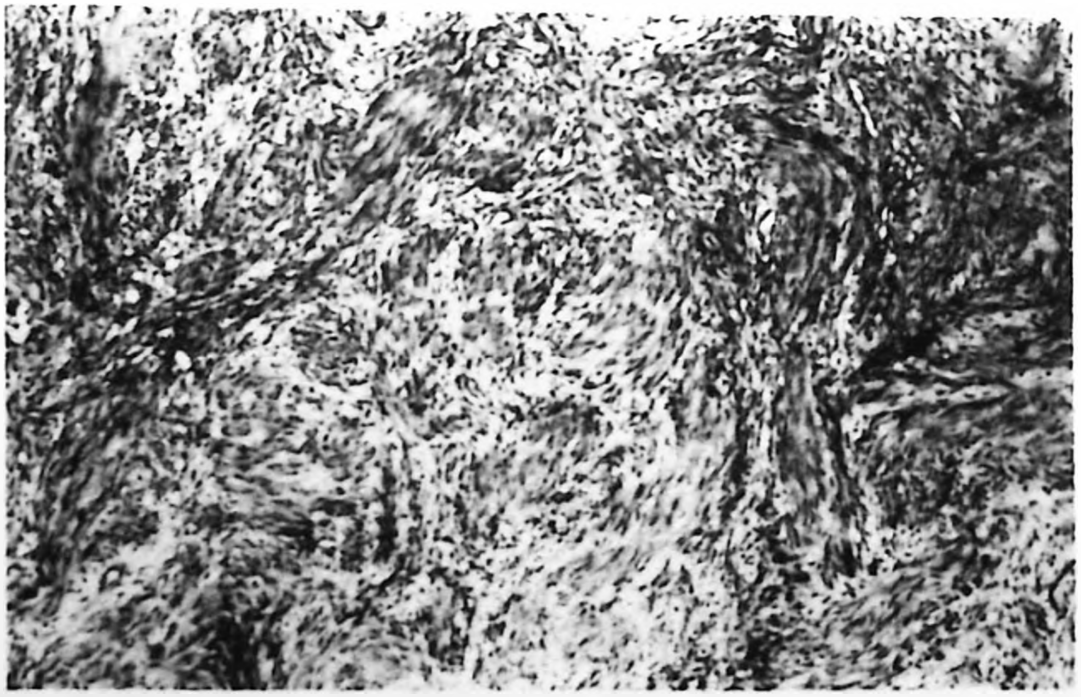


Fig. 1: Interlacing fascicles of spindle cells at the periphery of the tumor. (Trichrome stain, x 40).

In November 1991, a firm, paraspinal, subcutaneous 1.5 x 1.5 cm nodule was palpable in the left thoracolumbar area. X-ray and CT scan of this area were normal. Histologic evaluation of the excisional biopsy revealed hyalinized to hypercellular interdigitating bundles and nodules of spindle cells (Fig. 2). Mitotic figures were sparse. There were neither ossification and cartilage formations necrosis nor hemagiopericytoma-like areas. These findings were consistent with IM. The first tumor was also re-evaluated, and both tumors were found to be vimentin and a-smooth muscle actin-positive (Avidin-biotin peroxidase method with BioGenex Lab. antibodies). The diagnosis was revised as the multicentric form of IM. Chemotherapy was dicontinued. Skeletal radiograms, cranial CT and magnetic resonance imaging revealed no additional lesions. However, in October 1992, a skeletal survey revealed several osteolytic lesions on the skull and long bones of the extremities (Figs. 3, 4). Some of these lesions remained stable and some even regressed during the two-year follow-up period.

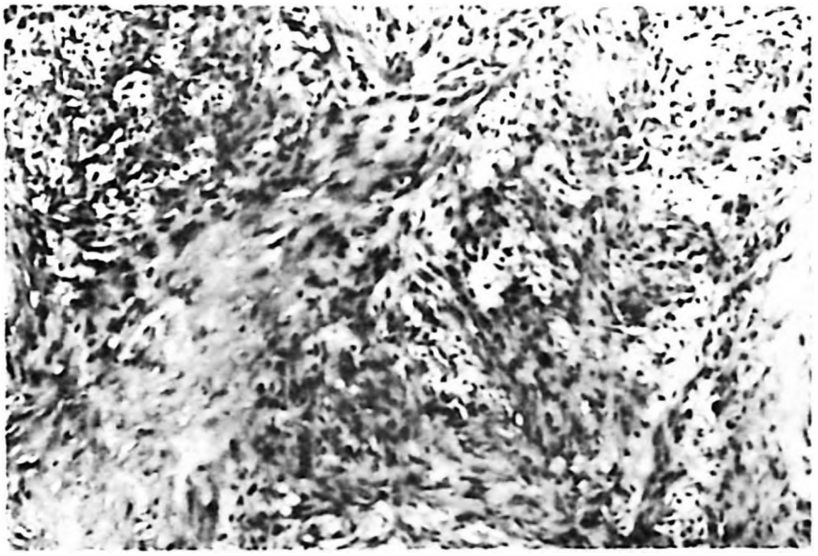
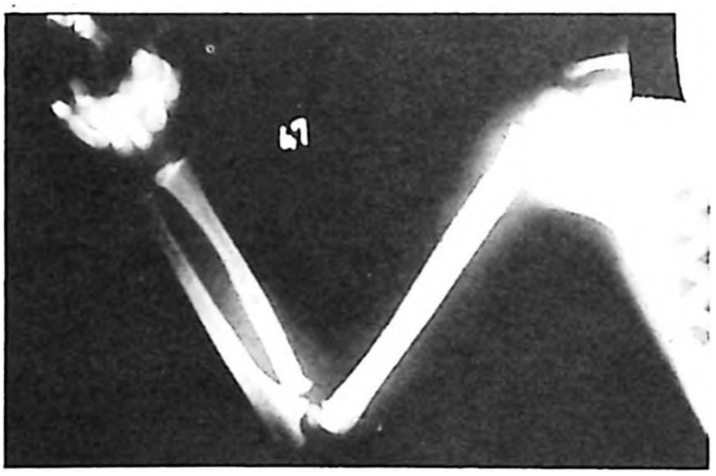
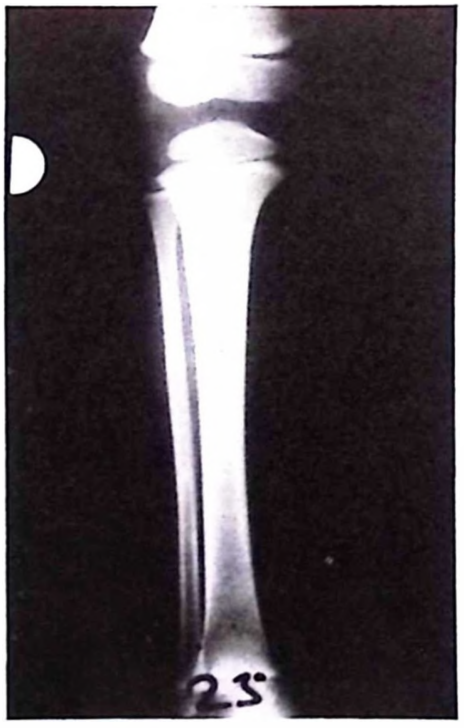


Fig. 2: Hypercellular, partially hyalinized interdigitating bundles of spindle cells. (H + E, x 100).

Fig. 3. Several osteolytic lesions of the skull



(a)



(b)

Fig. 4: Well-defined radioluscencies in the metadiaphyseal regions of the (a), radius and (b) tibia.

Discussion

Infantile myofibromatosis has formerly been described as multiple congenital mesenchymal hamartomas; fibromatoses; congenital diffuse, generalized hamartomatosis; congenital generalized fibromatosis and multiple vascular leiomyomas[1,2,4]. The diagnosis is established either at birth or during the first year of life[1,5,6]. A solitary, adult counterpart of this entity has also been described[7]. There is a male predominance of 1.6: 1[8,9].

The lesions cause few symptoms except for occasional pain caused by compression of nerves[8, 9]. In our case, facial paralysis was observed.

Although the cause of IM is unknown, its relation to estrogen hormones has been proven experimentally[8]. Both an autosomal-dominant[5] and an autosomal-recessive[10] pattern of inheritance have been described. Major malformations such as esophageal atresia and hypoplastic right kidney have been reported in an infant with IM[11].

There is still considerable debate as to the nature and histogenesis of this condition. It is probably of hamartomatous origin. The prominent cell type is the myofibroblast, which may exhibit a range of phenotypes including vimentin, α-smooth muscle actin, desmin and collagen type IV[1, 2, 12]. IM seems to display more advanced smooth-muscle differentiation than conventional fibromatoses[12, 13]. This differentiation was featured by α-smooth-muscle-actin positivity in our case.

IM occurs in either a solitary or multicentric form[1, 2, 4]. Chung and Enzinger[14] found solitary lesions to be at least twice as common as multiple lesions. Solitary lesions arise in subcutaneous tissue, muscle or bone. In the multicentric form, there are multiple soft-tissue, bone and visceral lesions[2]. Visceral lesions are not always associated with the multicentric form[14, 15]. If visceral lesions are present, the prognosis is poor[11, 16, 17]; otherwise the tumor follows a benign course, often with spontaneous regression[1, 9, 10]. Thus, conservative surgical treatment with local excision is recommended, but local recurrence has been documented in some cases[1, 6].

In the multicentric form, there may be a single nodule at the time of initial diagnosis, and formation of new nodules may be observed during the first few months of life[2, 8, 9, 18, 19]. New nodules are usually reported to appear within eight weeks, but in familial IM new nodules developed as late as four age[20]. Although there was no family history of similiar lesions in our case, a second subcutaneous nodule and multiple osteolytic lesions appeared 18 and 30 months after the initial nodule, respectively.

Bone involvement is represented as well-defined radioluscencies in the medulla of the craniofacial skeleton and metadiaphyseal regions of long bones[1, 2, 6, 7]. Therefore the radiological differential diagnosis of bone lesions includes Langerhans' cell histiocytosis, fibrous dysplasia, metaphyseal fibrous defect, aneurysmal bone cyst, chondromyxoid fibroma, familial multiple non-osteogenic fibromata, neurofibromatosis, metastatic neuroblastoma and hemangiomatosis[5, 6]. The histologic differential diagnosis presents greater difficulties, especially if the tumor is solitary. It must be differentiated from several soft tissue neoplasms, including fibrosarcoma, leiomyoma, leiomyosarcoma, hemangiopericytoma, neurofibroma and juvenile hyaline fibromatosis[5, 16]. Because of these difficulties, some cases have been misdiagnosed as sarcomas and given chemotherapy[11, 19].

Furthermore, chemotherapy has been initiated to prevent the rapid course of a multicentric form with visceral involvement[10] and based on the decision of the clinician in another case[20], but did not alter the clinical course in either.

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