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Infantile Myofibromatosis: A Case Report and Review of the Literature

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Infantile myofibromatosis is a rare mesenchymal disorder of infancy and childhood characterized by the formation of tumors in the soft tissues, muscle, bone, and viscera. Disease limited to the soft tissues, muscle, and bone has a good prognosis, and excision is curative; however, visceral involvement may be fatal. We present a case of infantile myofibromatosis in a 1-year-old boy and review the literature.

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Case Report

A 1-year-old Chinese boy presented with an asymptomatic erythematous nodule over the right palm that had been there since birth (Figure 1). The lesion was nontender and had been slowly increasing in size. There was no bleeding or ulceration. The patient was delivered by cesarean birth for fetal distress but was otherwise normal at birth. Developmental milestones were normal, and there was no family history of note. Clinically, there was a 1.2×1.0-cm nodule on the right palm that was firm and deep. The overlying skin was normal. There were no similar lesions elsewhere on the patient's body.

Results of a biopsy showed the lesion to be in the subcutis. There was a multilobular spindle cell proliferation in the subcutis that was surrounded by a compressed fibrous capsule. The spindle cells showed moderate pleomorphism and mitoses (Figure 2) and were arranged in storiform patterns and interlacing fascicles. There also was a prominent vascular component of small slitlike blood vessels with a tendency toward an angiocentric growth pattern, with satellite nodules at the periphery merging into the smooth muscle of blood vessel walls. Immunocytochemical analysis results showed the cells to be negative for desmin and to have a focal pattern of smooth muscle actin positivity (Figure 3). The histological features were those of infantile myofibromatosis.

Comment

Infantile myofibromatosis was first described in 1954 by Stout,[1] who termed the condition *congenital generalized fibromatosis*. In 1965, Kauffman and Stout[2] divided the condition into 2 categories: a multiple form limited to the skin, soft tissue, and bone, and a generalized form with visceral involvement. The solitary form of the disease was not appreciated until 1981, when Chung and Enzinger[3] recognized that the solitary presentation was more common than the multicentric form. Furthermore, they renamed the disease *infantile myofibromatosis* after observing morphologic characteristics of both smooth muscles and fibroblasts in the lesions. Wiswell et al[4] subclassified the condition into solitary and multiple lesions with further subdivision into the presence or absence of visceral involvement.

The disease commonly presents at birth or in infancy as solitary or multiple nodules or plaques ranging from 0.5 to 3 cm in diameter. Sixty percent of cases present at birth, and 90% occur within the first 2 years of life.[3] Lesions usually are in the skin, subcutaneous tissue, or muscle and may be found less frequently in the viscera and bone. The lesions generally are nontender and painless, and there may be associated ulceration. The solitary form is more common, affecting 70% of all cases and occurring predominantly in men. The most common sites of involvement for solitary lesions are the head and

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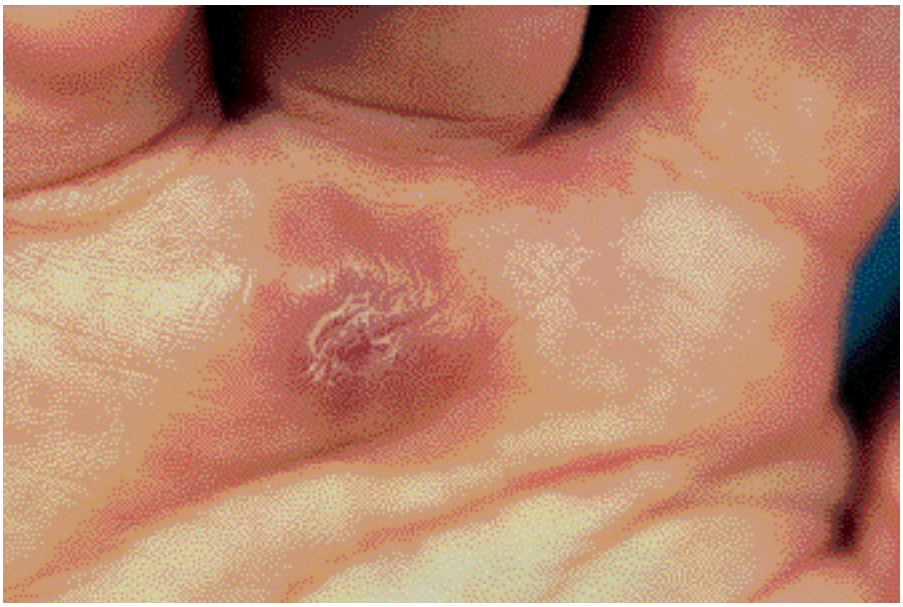

Figure 1. Erythematous nodule on the palm.

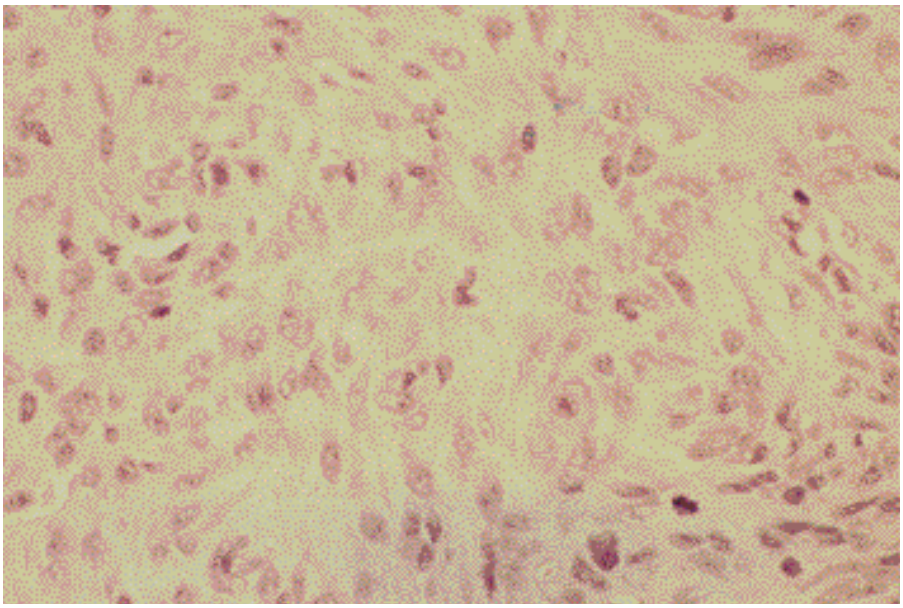

Figure 2. Tumor of spindle cells in the subcutis with pleomorphism and mitoses (right lower corner)(H&E, original magnification ×200).

neck, followed by the trunk and extremities.[3] Multicentric lesions appear more often at birth and in women.[3] The areas most commonly affected include the lungs, gastrointestinal tract, and myocardium. Familial cases are rare, but autosomal-dominant and recessive inheritances have been reported.[5-7]

Differential diagnoses include leiomyomas, hemangiopericytomas, congenital fibrosarcoma, metastatic neuroblastoma, and other infantile fibromatoses. Histologically, the tumor consists of cells intermediate between fibroblasts and smooth muscle cells arranged in fascicles or short bundles. There is a prominent vascular pattern, which is more centrally located and resembles hemangiopericytoma. Infantile myofibromatosis shares similar histological features with infantile hemangiopericytoma, and some authorities believe that these 2 conditions may represent a histological continuum with different stages of maturation of the same entity.[8] An origin from vascular subintimal mesenchymal or smooth muscle cells has been suggested[9] and is supported by the pathological findings in the present case.

In the absence of visceral involvement, the disease runs a benign, self-limiting course, with frequent spontaneous resolution and low recurrence rates

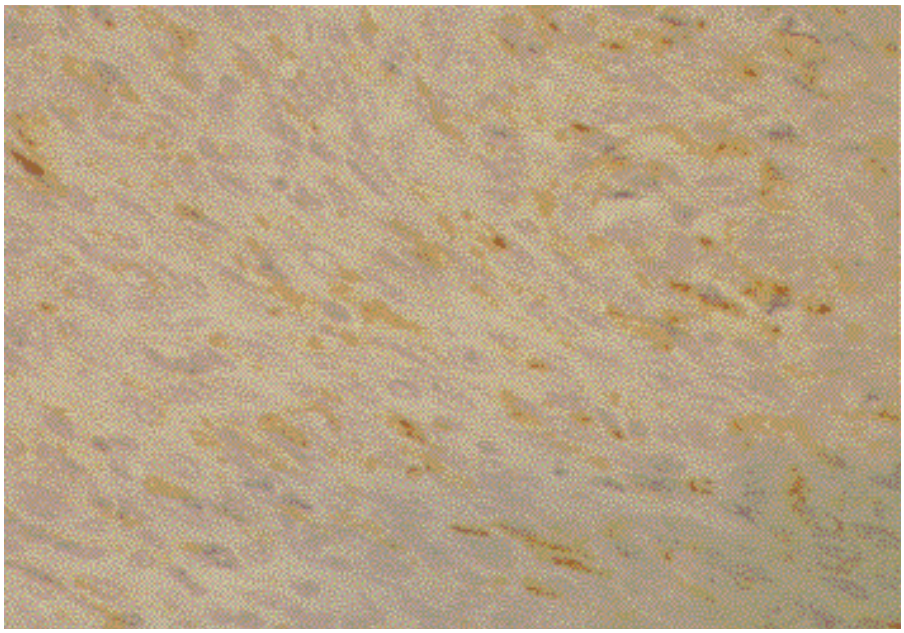

Figure 3. Immunocytochemical stain for smooth muscle actin shows patchy intracytoplasmic positivity (original magnification ×100).

after resection. Recurrence probably is due to inadequate removal of the tumor and can be cured by reexcision.[3] However, multicentric forms and the presence of visceral (particularly lung) involvement portends a poor prognosis.[4,10] According to Wiswell et al,[4] 73% of such patients died, mostly of cardiopulmonary failure. The evaluation of a patient with suspected infantile myofibromatosis should include a thorough family history, skeletal survey, chest x-ray, and computed tomography of the thorax and abdomen; in addition, a biopsy of the lesion should be performed.[11] Because lesions often regress spontaneously, surgical excision should be reserved for cases with involvement of vital structure.[11]

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