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Skeletal Radiology

Infantile myofibromatosis

Jean-François Chateil, M.D.[1], Muriel Brun, M.D.[1], Brigitte Lebail, M.D.[2], Yves Perel, M.D.[3], Jean-François Castell, M.D.[1], François Diard, M.D.[1]

[1] Unité de Radiopédiatrie, Hôpital Pellegrin-Enfants, Bordeaux, France
[2] Service d'Anatomie pathologique, Hôpital Pellegrin-Enfants, Bordeaux, France
[3] Service de Pédiatrie B, Hôpital Pellegrin-Enfants, Bordeaux, France

Case report

A 7-year-old girl was referred with a painless tumor in the posterior compartment of the left arm. The mass had appeared 6 months previously and had slowly grown. The child had no previous history of trauma.

Physical examination revealed a hard mass strongly attached to the left triceps muscle, measuring 6 cm in diameter. Plain film x-rays showed a posterolateral soft tissue tumor with numerous nodular or "cornflake" calcifications. The bone appeared intact and there was no periosteal reaction (Fig. 1). Sonography demonstrated a well-delineated lesion within the muscle, containing multiple calcifications (Fig. 2). A technetium-99m MDP scan revealed increased uptake in the soft tissue during the vascular and metabolic phases (Fig. 3). The rest of the skeleton was intact.

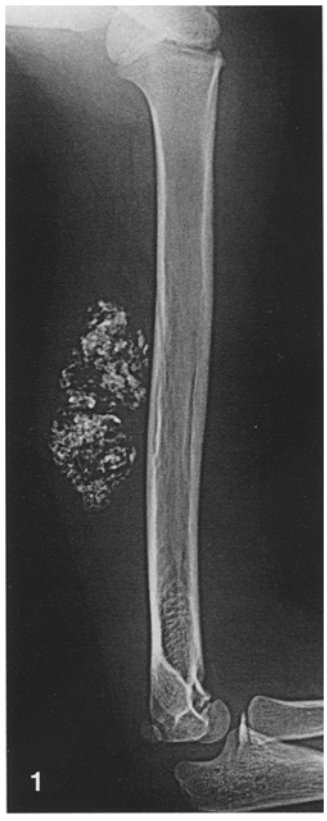
Computed tomography (CT) before and after intravenous contrast injection demonstrated an heterogeneous tumor close to the lateral head of the triceps muscle. The mass contained multiple central calcifications. The soft tissue component was hypodense compared to the muscle on plain CT and enhanced after the injection of intravenous contrast (Fig. 4). The adjacent bone was normal.

Correspondence to: Jean-François Chateil, M.D., Unité de Radiopédiatrie, Service de Radiologie A, Hôpital Pellegrin, Place A Raba Léon, F-33076 Bordeaux, Cedex, France

The girl underwent a local excision, and the mass was removed en bloc.

Macroscopically, the lesion was a well-delineated, nonencapsulated gray-white tumor measuring 6.5×3.0×1.5 cm. It was very firm on sectioning. The center was hard and fibrous and the histologic study required lengthy decalcification.

Microscopically, the lesion was circumscribed by thin collagen fibers. The tumor was composed of fusiform spindle cells arranged in fascicles with eosinophilic cytoplasm. Nuclei were small, ovoid, without atypia and showed no mitotic activity (Figs. 5, 6). The structure of the lesion was heterogeneous. There were numerous inflammatory cells with lymphocysts at the periphery, with fibrohyaline stroma, edema, foci of calcification and ossification,



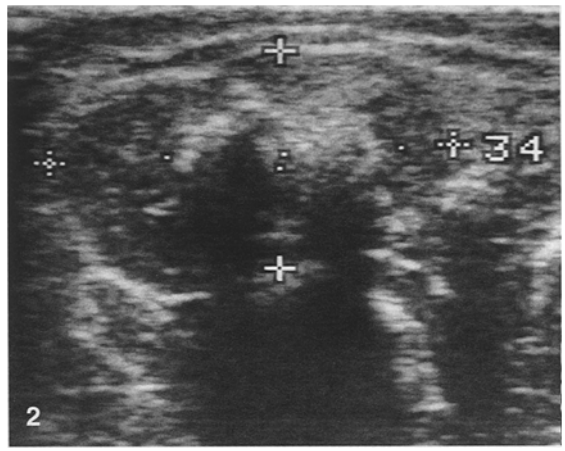


Fig. 1. Plain X-ray of the left arm shows a posterolateral soft tissue tumor with numerous "cornflake" calcifications. The adjacent bone is intact, without periosteal reaction

Fig. 2. Transverse sonogram shows a well-delineated lesion within the muscle, with numerous hyperechoic nodules

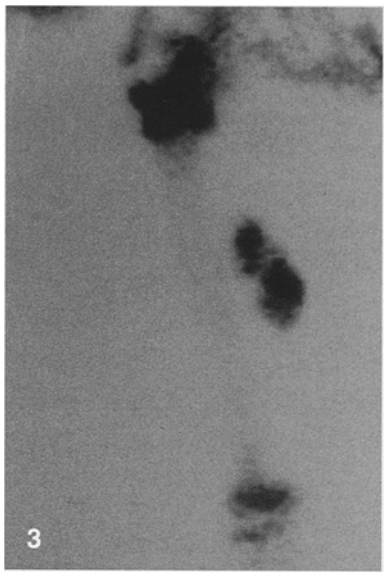


Fig. 3. Technetium-99m MDP bone scan of the left arm (metabolic phase with magnification) shows increased uptake in the tumor

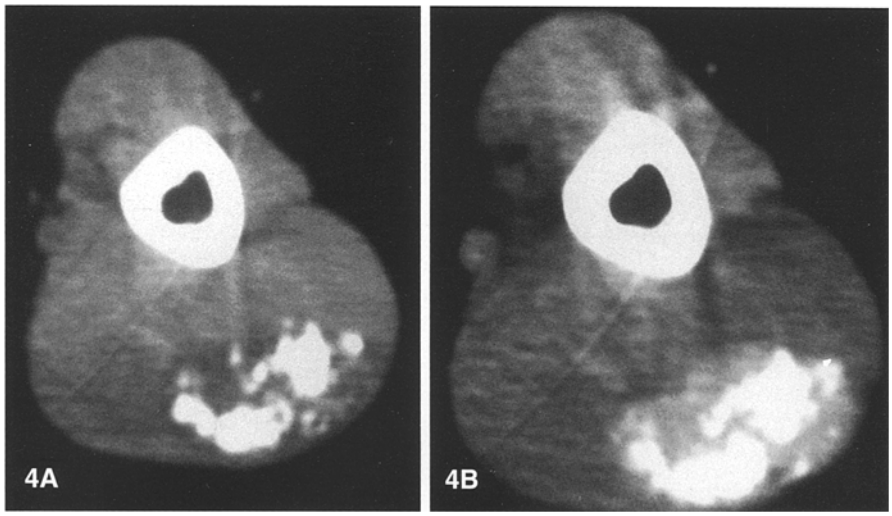


Fig. 4. Computed tomography (CT) scan. **A** Without contrast: the mass is centrally calcified; peripherical soft tissue component is hypodense compared to the muscle. **B** After intravenous contrast injection: there is a strong peripheral contrast enhancement

and a prominent vascular pattern at the center.

Immunocytochemical staining confirmed the mesenchymal origin of the tumor: the cells were strongly positive for vimentin and smooth-muscle actin. HHF 35 antibody was positive for most cells. One-third of them were positive for desmin. These findings strongly suggested a diagnosis of infantile myofibromatosis.

One year later, there was no evidence of recurrence, and the girl was clinically and radiologically well.

Discussion

Infantile myofibromatosis was first described by Stout, in 1954, as "congenital generalised fibromatosis". Several cases have been reported using different nomenclatures: congenital multiple or diffuse fibromatosis, multiple mesenchymal hamartomas, or multiple vascular leiomyomas of the newborn [1].

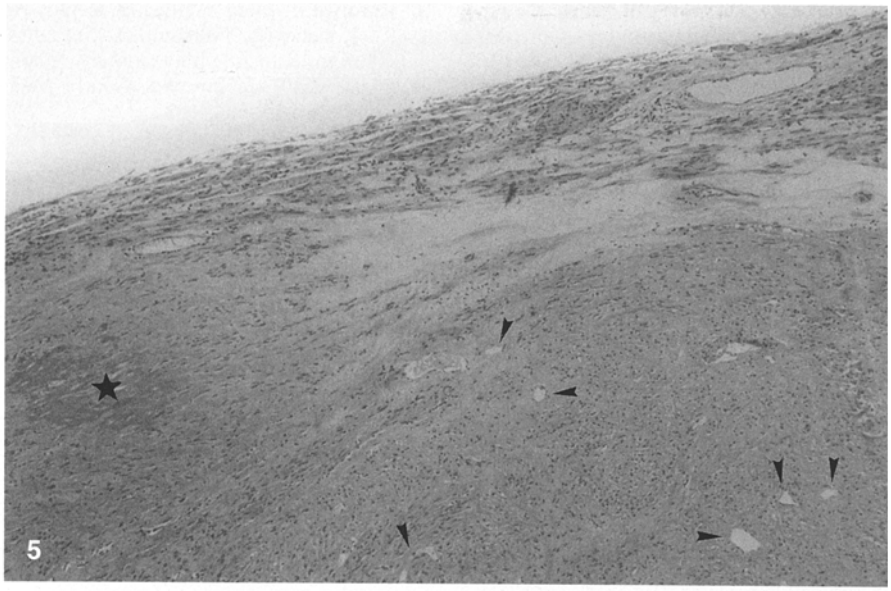
Chung and Enzinger [2], in 1981, revied 61 clinical and histological cases of congenital fibromatosis in children under 12 years and coined the term "infantile myofibromatosis." It is a mesenchymal disorder with nodular lesions within soft tissue, bone, or viscera. These tumors can be solitary or multiple, and can be rubbery, firm, or hard. Histological patterns are similar in all cases, with fusiform spindle cells and intermediate differentiation between fibroblasts and smooth muscular cells, called myofibroblasts. Calcifications may be present [2].

Infantile myofibromatosis mainly affects children under the age of 2 years (88% of cases) [2], but may develop later in infancy or in adults. Solitary and multicentric infantile myofibromatosis vary in their clinical aspects. Solitary infantile myofibromatosis (73% of cases) is mostly found in males (69%), and tends to be located in the head and neck (69%). Visceral location has occasionally been reported [3, 4] and bony involvement is also possible [5]. Multicentric infantile myofibromatosis (28% of cases) is mostly found in females (63%) and has a tendency towards soft tissue and bone involvement. Visceral lesions are common, often in multiple locations [4, 6, 7].

Prognosis depends on this visceral involvement: solitary and multicentric forms without visceral involvement have a good prognosis. In the multicentric forms, spontaneous regression is observed in 30–60% of cases [4]. Skin lesions usually remit spontaneously, sometimes leaving atrophic skin areas [8, 9].

The etiology of infantile myofibromatosis is unknown. Large doses of estrogens may produce multiple fibromas [7]. There is some evidence of genetic factors with hereditary transmission [2, 7]. Both autosomal recessive and autosomal dominant inheritance have been proposed [10].

Histological findings are typical in all cases. Macroscopically, the size of the tumor is variable (0.5–7.0 cm) [2]. The lesion is well defined, sometimes encapsulated, and is firm-to-hard on cutting. Microscopically, the tumor presents a multinodular aspect, each nodule has a zoned organization, with a central and a peripheral area [2, 11]. The central portion is made of small polygonal cells, with pale cytoplasm and basophilic, small round nuclei, corresponding to primitive cells. Normal mitosis occasionally takes place. There is often a prominent vascular pattern, having a characteristic hemangiopericytomatous appearance with thin-walled, arborizing and dilated vessels. The peripheral area of the nodule consists of fusiform spindle cells with eosinophilic cytoplasm, ovoid nuclei, well-defined cell margins, and tapered ends. These cells are arranged in in-



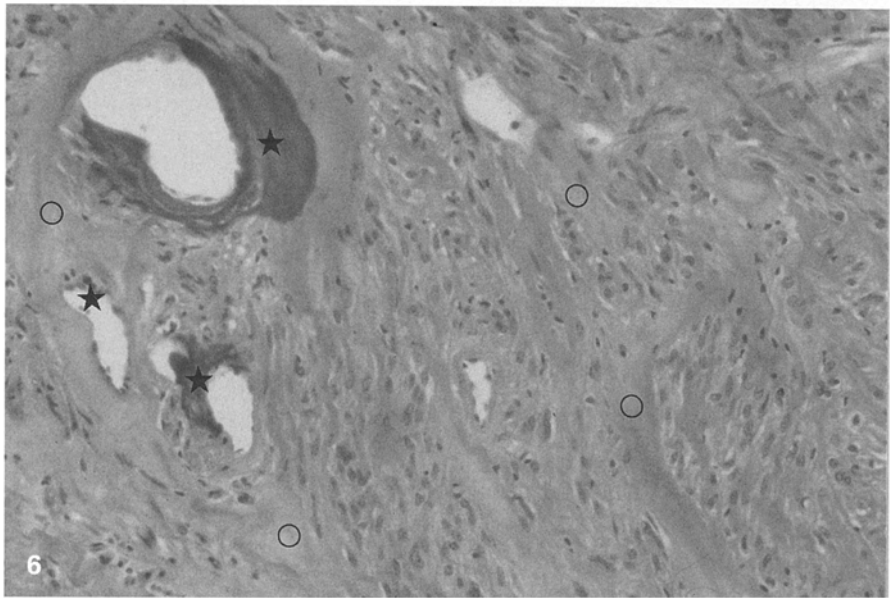


Fig. 5. Histological section (×45). The tumor is heterogeneous, peripherally well circumscribed with fusiform spindle cells. The center shows fibrohyaline stroma, foci of calcification (★) and a prominent vascular pattern (→)

Fig. 6. Histological section (×140). Fusiform small spindle cells arranged in short intertwining fascicles, with collagenous fibrosis (○) and calcifications (★). Nuclei are small, ovoid, and without atypia

terlacing or parallel fascicles. Occasionally the lesion shows, mainly in the center, foci of necrosis or hemorrhagic degeneration.

Irregular foci of calcification may be present [2]. Calcifications are unassociated with the necrosis. They are most often situated in the peripheral spindle cell areas or at the junction with the primitive cell areas [11]. In our case, calcifications were very numerous and located in all parts of the nodules.

Immunocytochemical staining is positive for vimentin and smooth-muscle actin, and negative for desmin [8, 12, 13], showing a mesenchymatous differentiation (vimentin positive), with intermediate cellular differentiation between the fibroblast and the smooth-muscle cells (actin positive and desmin negative). In our case, desmin staining was positive. The reason for this is controversial: desmin is a reliable indicator of myogenic differentiation, and staining results may be correlated with the proportion of fusiform cells.

Characteristic imaging features of infantile myofibromatosis in soft tissue locations vary. Plain x-rays almost always show a soft tissue tumor developed within muscle or subcutaneous tissue. Osseous scalloping may be present [14, 15]. Calcifications have been described, either with multiple foci within the lesion or as a peripheral rim [2, 15].

CT scans show a low-density region on a precontrast scan, with homogeneous enhancement after iodine injection. Locoregional extension is well demarcated, especially with soft tissue involvement [16–18]. Bone scans with Technetium-99m MDP may show increased extraosseous uptake in relation to the calcifications [15]. Magnetic resonance imaging may be helpful in defining the site and the extent of visceral lesions [19–21].

Possibilities to be included in radiological differential diagnosis of soft tissue lesions with calcifications include [15]:

1. Post-traumatic myositis ossificans, in which calcifications are almost always peripheral and the center radiolucent. The history of trauma may be difficult to assess.
2. Juvenile aponeurotic fibroma, which may be calcified, especially in the hands and feet.
3. Soft tissue chondroma, which shows frequently punctuated or nodular calcifications.
4. Soft tissue sarcoma (rhabdomyosarcoma, synoviosarcoma), which is calcified in 20% of cases.

In summary, an unusual case of infantile myofibromatosis with foci of calcifications was presented, with atypical age of onset and location. The radiological features of this tumor are not always evocative, and histological examination is necessary for diagnosis.

References

1. Boman F, Foliguet B, Metaizeau JP, Olive D, Rauber G. La myofibromatose infantile. Etude histopathologique et ultrastructurale d'une forme localisée à évolution spontanément régressive. Ann Pathol 1984; 4: 211–216.
2. Chung EB, Enzinger FM. Infantile myofibromatosis. Cancer 1981; 48: 1807–1818.

3. Waeltermann JM, Huntrakoon M, Beatty EC, Cibis GW. Congenital fibromatosis (myofibromatosis) of the orbit: a rare cause of proptosis at birth. Ann Ophthalmol 1988; 20: 394–399.
4. Wiswell TE, Davis J, Cunningham BE, Solenverger R, Thomas PJ. Infantile myofibromatosis: the most common fibrous tumor of infancy. J Pediatr Surg 1988; 4: 314–318.
5. Inwards CY, Unni KK, Beabout JW, Shives TC. Solitary congenital fibromatosis (infantile myofibromatosis) of bone. Am J Surg Pathol 1991; 15: 935–941.
6. Goldberg NS, Bauer BS, Kraus H, Crussi FG, Esterly NB. Infantile myofibromatosis: a review of clinicopathology with perspectives on new treatment choices. Pediatr Dermatol 1988; 5: 37–46.
7. Salamah MM, Hammoudi SM, Sadi ARM. Infantile myofibromatosis. J Pediatr Surg 1988; 23: 975–977.
8. Bellman B, Wooming G, Landsman L, Penneys N, Schachner LA. Infantile myofibromatosis: a case report. Pediatr Dermatol 1991; 8: 306–309.
9. Parker RK, Mallory SB, Baker GF. Infantile myofibromatosis. Pediatr Dermatol 1991; 8: 129–132.
10. Bracko M, Cindro L, Golouh, R. Familial occurrence of infantile myofibromatosis. Cancer 1992; 69: 1294–1299.
11. Fletcher CDM, Achu P, Van Noorden S, McKee PH. Infantile myofibromatosis: a light microscopic, histochemical and immunohistochemical study suggesting true smooth muscle differenciation. Histopathology 1987; 11: 245–258.
12. Speight PM, Dayan D, Fletcher CDM. Adult and infantile myofibromatosis: a report of three cases affecting the oral cavity. J Oral Pathol Med 1991; 20: 380–384.
13. Vigneswaran N, Boyd DL, Waldron CA. Solitary infantile myofibromatosis of the mandible. Oral Surg Oral Med Oral Pathol 1992; 73: 84–88.
14. Kozlowski K, Kan A, Marsden FW. Case report 684. Skeletal Radiol 1991; 20: 397–399.
15. Soper JR, De Silva M. Infantile myofibromatosis: a radiological review. Pediatr Radiol 1993; 23: 189–194.
16. Kuroiwa T, Ohta T, Kazuki S, Ogawa R, Kobata H, Tsutsumi A. Infantile myofibromatosis with a solitary lesion in the skull. Case report. Neurol Med Chir 1990; 30: 184–187.
17. Hutchinson L, Sismanis A, Ward J, Price L. Infantile myofibromatosis of the temporal bone: a case report. Am J Otol 1991; 12: 64–66.
18. Stautz CC. CT of infantile myofibromatosis of the orbit with intracranial involvement: a case report. AJNR 1991; 12: 184–185.
19. Moore JB, Waldenmaier N, Potchen EJ. Congenital generalized fibromatosis: a new management strategy provided by magnetic resonance imaging. AJDC 1987; 141: 714–716.
20. Stautz CC. Magnetic resonance evaluation of infantile myofibromatosis. Mo Med 1990; 87: 889–891.
21. Wiswell TE. Infantile myofibromatosis and the use of magnetic resonance imaging. AJDC 1988; 142: 486.