Radiology Case Reports 19 (2024) 21–23

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Case Report

Solitary infantile myofibromatosis in the upper extremities: Case report ☆,☆☆

Amsiguine Najwa[a,*], *Izi Zineb*[a], *El Ghaffouli Sara*[b], *Rouas Lamiae*[b], *Lrhorfi Najlaa*[a], *El Haddad Siham*[a], *Allali Nazik*[a], *Chat Latifa*[a]

[a] *Department of Pediatric Radiology, Faculty of Medicine, Hospital Ibn Sina, Rabat, Morocco*
[b] *Department of Pediatric Anatomopathology, Faculty of Medicine, Hospital Ibn Sina, Rabat, Morocco*

ARTICLE INFO

Article history:
Received 17 June 2023
Revised 13 September 2023
Accepted 20 September 2023
Available online 28 October 2023

Keywords:
MRI
Myofibroma
Solitary
Upper limbs

ABSTRACT

Infantile myofibromatosis (IM) is a mesenchymal tumor that may present in infants in a couple of major forms: solitary (myofibroma) and multicentric (myofibromatosis) which can be more subdivided into IM without or with visceral involvement. The tumors present as nodular lesions in the soft tissues, bones, and/or internal organs. Although the success of imaging in suggesting the correct diagnosis can't be denied, histopathology and Immunohistochemical examinations are necessary to confirm the diagnosis of IM as it might be misdiagnosed as a malignant tumor. We report a case of solitary infantile myofibromatosis in the upper extremities discovered in a 9-year-old girl. She had swelling and an enlargement on the posterior forearm on the ulnar side. The X-ray showed a lytic lesion with swollen soft tissue. The patient underwent an MRI which suggested the diagnosis of myofibroma. Then, solitary myofibroma was confirmed histologically. Infancy's most prevalent fibrous tumor is IM. Its prognosis depends on the visceral involvement. Imaging, especially MRI is the ideal tool to diagnose it.

Introduction

The World Health Organization (WHO) characterized myofibromatosis as a benign fibroblastic-myofibroblastic lesion in its 2002 classification of soft tissue lesions. This syndrome often manifests before the age of 2, although it can also be seen in older kids and even adults. The distribution primarily affects the head, neck, and torso, with only a very small percentage of cases involving the limbs [1]. Patients with lesions of viscera might have a poor prognosis and high mortality. Although it was reported that the disease was hereditary and the inherited genes related to autosomal or genetic heterogeneity rather than sex-limited chromosomes might play an important role, the etiology of IM still remains unclear [2]. The present study describes the imaging modalities via a case of

☆ Acknowledgments: Not applicable.
☆☆ Competing Interests: The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.
* Corresponding author.
E-mail address: malaklina.zark@gmail.com (A. Najwa).
https://doi.org/10.1016/j.radcr.2023.09.063

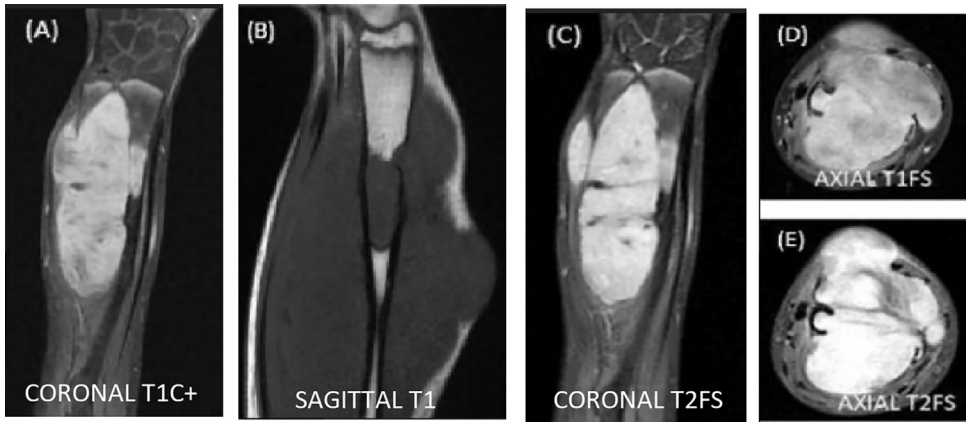

Fig. 1 – MRI of the upper right forearm showing a tumoral process developing on bone extending to soft tissues, isointense in T1-WI in sagittal slice (B) compared to the muscle, hyperintense in T2-WI (C and E) with marked enhancement after injection of Gadolinium (A and D). The aggressive tumor is responsible for the lysis of the cortical bone with the invasion of the medulla.

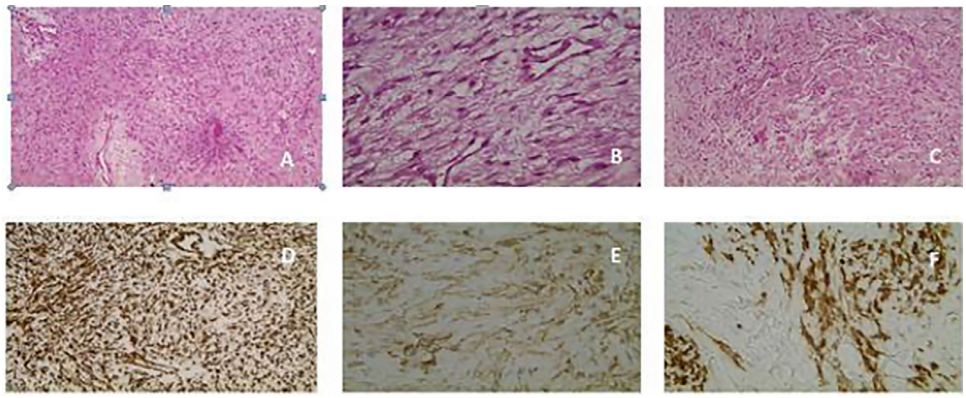

Fig. 2 – (A) Proliferation of myofibroblastic cells on a loose myxoid background. Some vessels have a hemangiopericytic appearance (lower left). Hematoxylin eosin, x 20 (B) Myofibroblastic cells have ovoid or elongated nuclei and do not show severe atypia. Fine capillary vessels are compressed by this proliferation. Hematoxylin eosin, x 40(C) keloid-like collagen foci. HE, x 40 (D) Tumor cells are positive with vimentin Immunohistochemistry, x 20 (E) Tumor cells show positive cytoplasmic labeling with beta-catenin. Immunohistochemistry, x 40 (F) Tumor cells are smooth muscle actin positive.

solitary IM involving soft tissues of the upper extremities in females who were 9 years old. The case is unusual in its symptom presentation and the tumor's origin is rarely observed.

Case report

A 9–year–old female patient was admitted to the Children's hospital of Ibn Sina with a few weeks' history of growth in her right forearm. Otherwise healthy, the girl had swelling and a 4 × 5 cm enlargement on the posterior forearm on the ulnar side. No relevant family history was noted. X-rays showed no fractures but swollen soft tissue. The patient underwent an MRI (Fig. 1), where the diagnosis of solitary myofibroma was first introduced and further confirmed histologically (Fig. 2). The postoperative course was uneventful with a full resection of the nodule up to 0.5 cm above the margins. In the following year, no recurrence was identified either locally or systemically.

Discussion

In children and infants, IM is thought to be the most typical fibrous tumor. Skin, subcutaneous tissue, internal organs, or bones may develop one or more nodular lesions, which are the hallmark of IM. Similar histological findings are found in both the solitary and multicentric forms, although the clinical characteristics and prognoses differ. Exclusive soft tissue lesions are seldom observed with the solitary type. The majority of the bone's solitary IM instances have affected the craniofacial bones [1]. The incidence of soft tissue tumors in newborns is about 1/15,000 and 35% of them were IM [2].

The exact etiology remains unknown but given the recent advances in understanding the molecular pathology of myofibromas, all patients, regardless of family history, stand to benefit from genetic testing both of their germline and tumor tissue for mutations in PDGFRB [3]. Both autosomal dominant and recessive transmission have been proposed, but no genetic basis has been proven [4].

Histologic investigation of tumor nodules reveals cells' intermediate appearance between fibroblasts and smooth muscle cells. As in this case, a highly vascular central region resembling a hemangiopericytoma is commonly observed (Fig. 2). Recent reports have proposed a histogenic relationship or spectrum of disease relating to IM and infantile hemangiopericytoma, questioning a true distinction between the 2 entities [4].

The disease in this case can be frequently mistaken during diagnosis for nerve sheath tumors, intramuscular myxoma, intramuscular lipoma, synovial sarcoma, malignant peripheral nerve sheath tumor, muscle metastasis, primary muscle tumors, or other soft tissue sarcomas [5].

Radiographic findings of these neoplasms vary according to the involved tissue type. Extraosseous lesions may be unrevealing or may demonstrate a soft tissue mass with dystrophic internal calcifications. Bone involvement may present as well-circumscribed, eccentric, metaphyseal, and lytic lesions. Calcification and border sclerosis may develop in mature lesions, while periosteal reaction or cortical erosion may be present in earlier lesions. Pathologic fractures have been described [5].

MRI is the ideal imaging tool to further characterize soft tissue abnormalities, it can be useful in determining the myofibromatosis's extent, course, and prognosis. However, MRI findings of myofibroma are mostly case reports, so more studies have to be made to better characterize this entity. As in our case on T1WI, tumors appear iso/hypointense relative to the adjacent skeletal muscle. On T2WI and FS T2WI, the tumor is heterogeneous with hyperintensity relative to the adjacent skeletal muscle, but a relatively lower signal to the fat. On FS contrast-enhanced T1WI, the tumors showed marked enhancement, but with irregular strips or/and patchy hypointensities [6] (Fig. 1).

Most IM solitary or multicentric regress spontaneously, though their number may increase, and there are also reports of aggressive recurrences. Therefore, we conclude that this condition has an excellent prognosis globally if visceral involvement is absent, in the opposite case the mortality has been estimated at up to 76%. The management of IM in children starts with the confirmation of the diagnosis with a biopsy and then the search for evidence of visceral involvements using imaging specially MRI.

Surgical excision is the treatment of choice in solitary forms, which was the case for our patient. Otherwise, regular follow-up alone is warranted. Modalities of treatment other than surgery are reserved for such lesions and for symptomatic yet nonresectable tumors. These include radiotherapy, local glucocorticoid injections, and chemotherapy using vincristine, actinomycin D, and cyclophosphamide [7].

Conclusion

IM should be included in the differential diagnosis of an infant or child with solitary or multiple nodules. The presence of visceral involvement affects the prognosis. The role of imaging is crucial for the delineation of size, location, extent, and effect on adjacent structures although the magnetic resonance imaging findings are remarkably consistent, they tend to be nonspecific.

Patient consent

Written informed consent was obtained from the patient for the publication of this article

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