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A misleading tumor: A rare form of infantile fibromatosis

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Abstract

Infantile fibromatosis is a rare mesenchymal disorder characterized by the fibrous proliferation of the skin, bone, muscle, and viscera. The clinical features vary from solitary to multicentric forms with similar pathological features. Although the tumor is histologically benign, it is a highly infiltrating lesion making the prognosis poor for patients with craniofacial involvement affection due to the major risk of nerve vascular and airway compression syndrome. The solitary form of infantile fibromatosis observed in the dermis, subcutis, or fibromatosis tends to occur predominantly in males and typically affects craniofacial deep soft tissues. We present a case of an unusual symptom presentation and a rarely observed location of a solitary fibromatosis form, affecting the muscle of the forearm and infiltrating the bone in a 12-year-old girl. Imaging findings were suggestive of rhabdomyosarcoma, but histopathology set the diagnosis of an infantile fibromatosis. The patient, then, received chemotherapy, and amputation was proposed due to the inextricability of this benign yet aggressive tumor, an option that was refused by her parents. We discuss through this article the clinical, radiological, and pathological features of this benign yet aggressive condition, the potential differential diagnosis, the prognosis, and treatment options substantiated with concrete examples from the literature.

Keywords

Infantile fibromatosis, mesenchymal tumors, magnetic resonance imaging, histopathology, tumor

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Introduction

The term fibromatosis describes an entity of mesenchymal disorder with a fibrous proliferation of the skin, bone, muscle, and viscera.[1]

Although rare, it is the most common fibrous tumor in childhood.[2]

It encompasses a spectrum from benign fibrous lesions to fibrosarcomas, with both superficial and deep types. The deep variant presents an aggressive diffuse, multicentric, or solitary infiltration similar to malignant lesions, with alternating periods of rapid and arrested growth.[3]

Spontaneous involution is common, and there has been no report of any case of sarcomatous degeneration.[4]

The solitary type has a better prognosis than those with multicentric lesions.[5]

Case

We report the case of a 12-year-old girl presented with a swollen right forearm, painful on palpation gradually increasing in volume.

X-ray examination exhibited a soft-tissue mass encompassing the two bones of the forearm in a circumferential way, with well-defined margins, associated with medullary bone lysis, a significant discontinuous spiculated periosteal reaction, and a deformation of the bones' axes (Figure 1(a)). Ultrasound showed a heterogeneous poorly vascularized mass (Figure 1(b)).

Magnetic resonance imaging (MRI) was then performed for a better assessment demonstrating a homogeneously isointense mass involving both superficial and deep soft-tissue layers on T1w, deforming the bone with a high heterogeneous signal on T2w, avidly enhancing after gadolinium administration.

The medullar bone lesion presents similar intensity changes as the soft-tissue mass (Figure 2).

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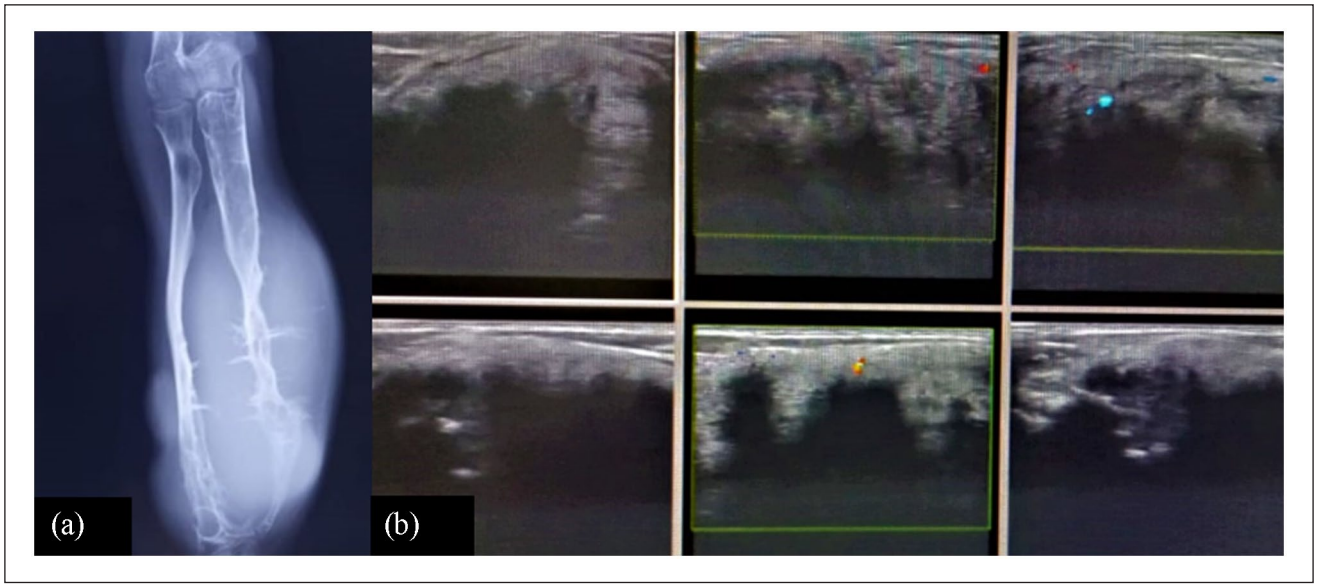

Figure 1. X-ray (a) demonstrates a well-circumscribed osteolytic lesion involving the diaphysis of both the right radius and ulna with spiculated periosteal reaction. Ultrasonography images (b) show a heterogeneous intra-muscular mass, containing hypoechoic areas, and poorly vascularized on color Doppler.

These findings arouse suspicion of a rhabdomyosarcoma.

A biopsy was performed, and surprisingly, histopathology revealed that the patient had infantile fibromatosis.

The patient received three cycles of vincristine-, actinomycin D-, and cyclophosphamide-based chemotherapy to shrink the tumor and stop its progression, but there was no response. Due to the high degree of infiltration and involvement of the vascular and nervous structures, partial excision was not possible. As a result, a radical surgery with forearm amputation was proposed, which the parents refused.

Discussion

Infantile desmoid fibromatosis is a rare, benign tumor that infiltrates soft tissues that was first reported in children by Stout in 1954 and called "generalized congenital fibromatosis," and in 1981, Chung and Enzinger[6] proposed the term "myofibromatosis infantile."

Fibromatosis is formed by benign fibroblastic tumors that develop multicentrically and independently from the organ's different fibroblastic contingents. The prevalence is estimated at 1/150,000 live births.

There are two forms of the disease, a solitary form and a disseminated form with multiple lesions. The second, less common form, may be accompanied by visceral involvement, which can be fatal. Lesions in children are most often present at birth in 60% of cases or appear during the first 2 years of life in 88% of cases.[5] We distinguish two forms of the disease; the solitary form which is more common in male patients (69%) and affects mainly the deep soft tissues of the head-neck region and the trunk. The multicentric form, on the contrary, predominated in female patients (63%), which was found not only in soft tissues but in bones and viscera as well.[6]

Autosomal dominant transmission with variable penetrance has been proposed due to familial cases reported in previous observations.[7]

Two causative genes have been identified: PDGFRB and NOTCH3 encoding the PDGFRB and NOTCH3 proteins, respectively. PDGFRB is a tyrosine kinase receptor for platelet-derived growth factors that are mitogenic for cells of mesenchymal origin. PDGFRB expression is upregulated by NOTCH3, suggesting that genetic abnormalities in both genes are involved in the same mechanism.[8]

The morphological examination shows a well-defined, deep dermal, and hypodermic nodule, consisting of a biphasic cell proliferation with a zone phenomenon. In the periphery, there is a proliferation of spindle-shaped cells with abundant eosinophilic cytoplasm arranged in bundles, of myoid differentiation. Sometimes these cells form buds in the vascular lumens from which they remain separated by the endothelial lining. In the center, the spindle-shaped cells of smaller size are arranged in bundles of concentric arrangement around branched vessels creating a hemangiopericytic appearance. This central area could be necrotic, hemorrhagic, or calcified.[9]

Both cellular components express alpha-smooth muscle actin (alpha-SMA) and vimentin on immunohistochemistry and are negative for desmin and protein S100.[10]

Myofibromas are usually characterized by heterogeneous density or signal intensity, with moderate or marked enhancement and irregular non-enhancing hypointense strips compatible with fibrous zones. These imaging

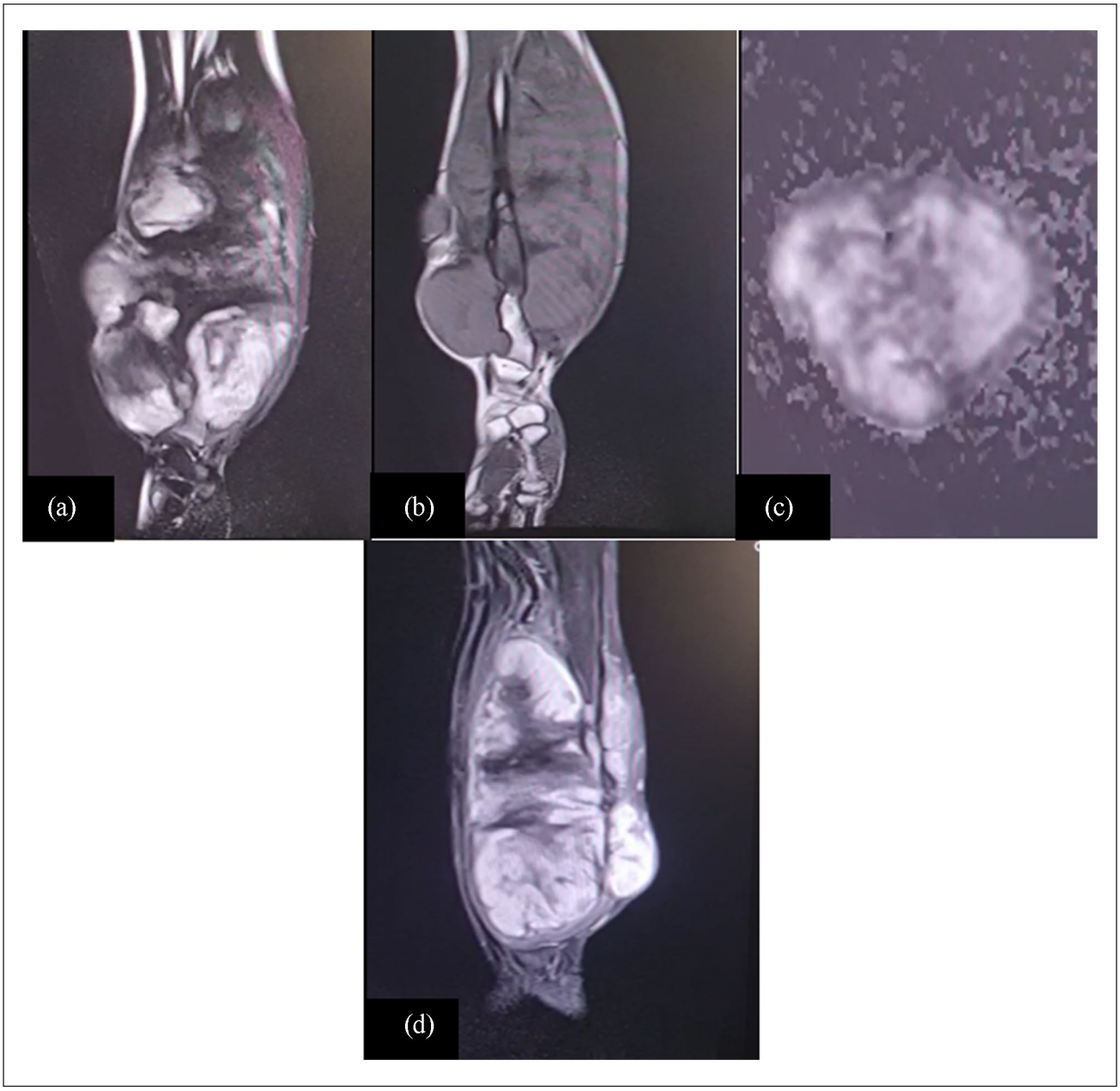

Figure 2. A 12-year-old young girl with infantile fibromatosis in the right forearm. (a) Coronal T2WI demonstrates a lobulated, heterogeneous hyperintense mass with irregular strip hypo intensities (arrows). (b) On coronal T1WI, the mass is homogeneous and isointense, with medullar bone lesion presenting the same characteristics. (c) The lesion is hyperintense on diffusion. (d) On contrast-enhanced axial FS T1WI, the lesion shows inhomogeneous marked enhancement with non-enhanced hypointense fibrous areas.

characteristics are non-specific of fibromatosis, and the final diagnosis must be made by pathology. Yet, it may be helpful in the diagnosis of tumors and in differentiating them from other bone and soft-tissue tumors on imaging. Aggressive fibromatosis shows paradoxical signal intensity in MRI. It presents both long and short T2 due to its significant fibrous elements and marked hypocellularity. This imaging characteristic may be seen in both benign and malignant soft-tissue lesions. Histologic composition of the tumor rather than the histologic diagnosis appears to influence the magnetic resonance (MR) signal on T2-weighted sequences as studies showed.[11]

In the differential diagnosis, congenital or infantile desmoids-type fibromatosis, fibrosarcoma, fibrous histocytoma, leiomyosarcoma, neurofibromas, osteoblasts mass, and fibrous dysplasia must be considered. All those lesions have specific histological, immunohistochemical and ultrastructural features, and different natural histories.

The prognosis of fibromatosis varies according to the type and localization. Forms without visceral involvement

have an excellent prognosis, with a spontaneous regression of the lesions in 1–2 years. Generally, the prognosis of solitary musculoskeletal fibromatosis is favorable with persistence risk of recurrence.

Treatments with no long-term effect are preferred due to the benign nature of the lesions. For lesions affecting the skin and/or muscles, treatment is not recommended and a monitoring protocol is proposed due to the tendency to spontaneous regression. Radical surgical excision is necessary if the lesions are located in areas at risk or if they are symptomatic. In case of incomplete resection, a new excision can be proposed later due to the risk of recurrency. The basic treatment is based on the weekly administration of methotrexate and vinblastine in low doses. It is indicated in case of progressive multifocal lesions involving the vital prognosis.[1,2] Other treatments, such as conventional chemotherapy (vincristine, actinomycin D, and cyclophosphamide), should be reserved for patients with rapid symptomatic progression due to the long-term risk of secondary malignancies. PDGFRB inhibitors have not yet been evaluated for the treatment of this disease.[12]

Conclusion

Solitary localization in forearm muscles of an infantile fibromatosis is very rare, especially in a 12-year-old female, manifesting as a swelling-isolated musculoskeletal mass mimicking malignant tumor partly rhabdomyosarcoma.

Ultrasound, computed tomography, and MRI do not allow differentiating the diverse types of fibrous lesions, but are very essential, showing the localization, and the infiltrated tissues. Histology is the only tool to make the definite diagnosis.

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Authors contributions

Ibtissam El Ouali: Conception of the work, design of the work and acquisition of data.
Kenza Berrada: Acquisition of data.
Ibrahima Diallo Dokal: Acquisition of data.
Rachida Saouab: Drafting the work.
Jamal El Fenni: Revising the work critically for important intellectual content.
Tariq Salaheddine: Revising the work critically for important intellectual content and final approval of the version to be published.

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Ethics approval

Our institution does not require ethical approval for reporting individual cases or case series.

Informed consent

Written informed consent was obtained from a legally authorized representative(s) for anonymized patient information to be published in this article.

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