

Mazabraud Syndrome: Clinical Review and Therapeutic Approach Regarding a Case Report

Síndrome de Mazabraud: Revisão Clínica e Abordagem Terapêutica de um Caso Clínico

Keywords: Fibrous Dysplasia of Bone/diagnostic imaging; Magnetic Resonance Imaging; Myxoma

Palavras-chave: Displasia Fibrosa Óssea/diagnóstico por imagem; Mixoma; Ressonância Magnética

Dear Editor,

Mazabraud syndrome is a rare condition, often undiagnosed, predominantly affecting women (68% of the cases) in their forties.¹ This syndrome is characterized by the association of intramuscular myxoma (IM) and benign soft tissue tumors. The latter are typically painless and slow-growing, primarily found in the leg muscles, especially the

quadriceps, either solitary or multiple, with fibrous dysplasia (FD), defined as a benign fibro-osseous lesion that replaces healthy bone. There is no consensus regarding the incidence of monostotic (FD in one bone) and polyostotic (FD in several bones) forms.^{1,2}

We describe the case of a 63-year-old woman who presented with tumors in the right thigh for the past five years that had been increasing in size and causing pain, as well as limiting the movement of her right lower limb. On physical examination, there were masses that were painless and had a hard consistency on her right thigh, there was limitation in the movement of her right lower limb and difficulty walking, which she reported had been worsening over the past few months. A magnetic resonance imaging (MRI) scan detected FD in the femur and multiple IMs (Fig. 1). A biopsy confirmed a diagnosis of Mazabraud syndrome.

Soft tissue IMs, which appear around the sixth decade

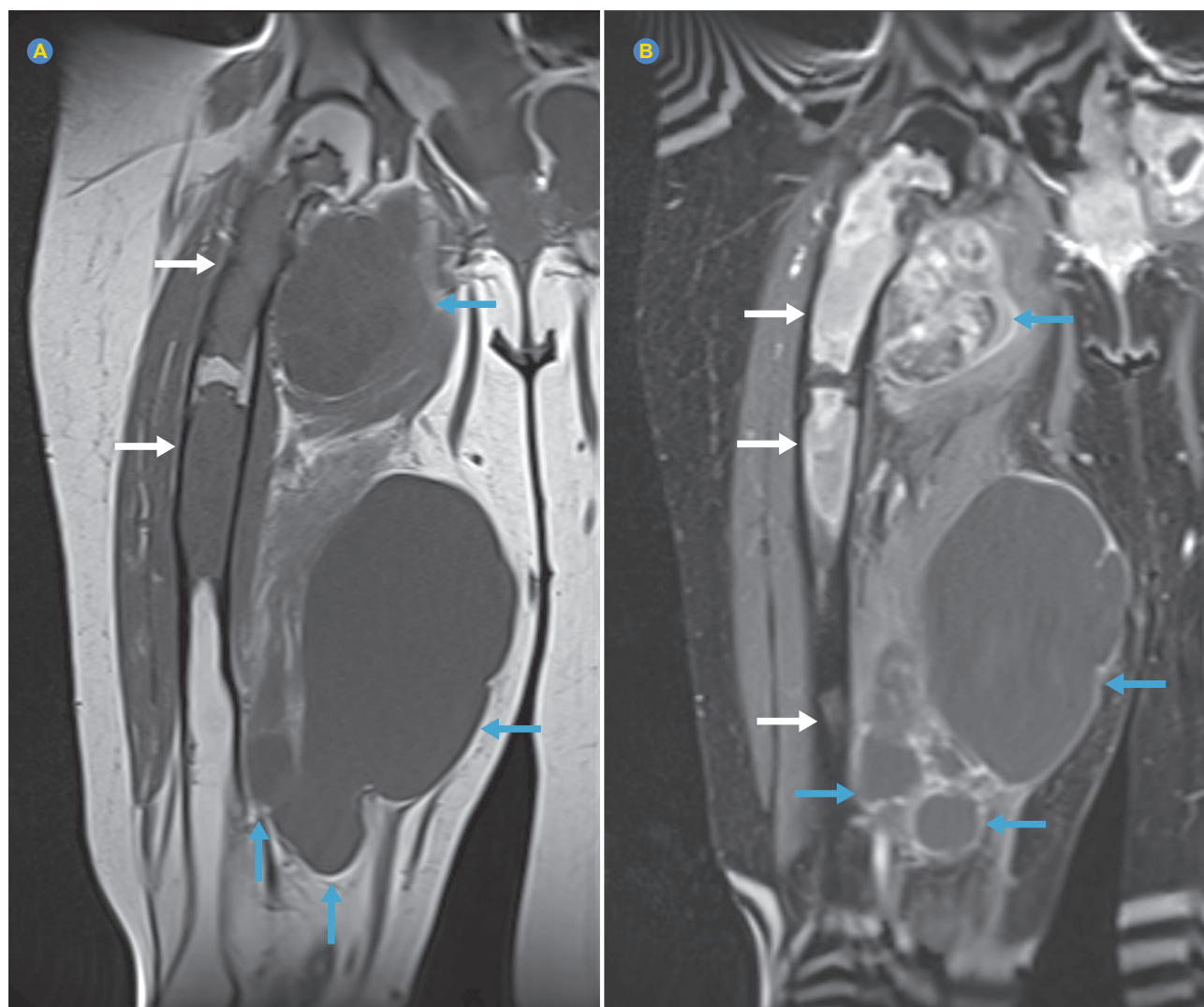


Figure 1 – T1 weighted-image MRI in coronal section (A); T1 FAT SAT DIXON with contrast weighted-image MRI in coronal section (B). MRI revealed extensive heterogeneity of the femur with an expansile appearance, significantly affecting its proximal portion, with heterogeneous contrast enhancement, in a 'cane-shaped' configuration, consistent with fibrous dysplasia (white arrows). Multiple lobulated intramuscular images with peripheral contrast enhancement and others with heterogeneous contrast enhancement, consistent with intramuscular myxomas (blue arrows).

of life and have a benign course, are considered soft tissue hamartomas with tumor-like growth.^{3,4} Furthermore, it is important to note that the development of FD usually precedes the appearance of IMs by approximately 6.5 years. Sarcomas can develop in areas of FD, as they have a rare but clear potential for malignant transformation. The most common malignancies that can arise from FD are osteosarcoma (70%), followed by fibrosarcoma (20%), and chondrosarcoma (10%).^{1,2}

Therapeutic management varies from clinical surveillance and imaging evaluation to orthopedic surgical intervention in symptomatic patients. Prolonged use of bone antiresorptive agents, like zoledronate, may be considered a viable alternative. These agents appear to reduce the volume of myxomas; however, they do not demonstrate significant effects on the regression of underlying FD.⁵ Treatment options for Mazabraud syndrome include extensive excision of myxomas, particularly when painful or compressive symptoms are present. It should be emphasized that even after partial excision of myxomas, the possibility of local recurrence persists, underscoring the need for regular follow-up to detect local recurrences and the emergence of new lesions. The therapeutic approach to FD, in general, includes bisphosphonate therapy, with surgical intervention rarely being necessary. The concurrent presence of myxomas requires closer monitoring of FD lesions due to the potential risk of malignant transformation.⁵

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Recebido/Received: 24/09/2023 - **Aceite/Accepted:** 16/01/2024 - **Publicado/Published:** 01/04/2024

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<https://doi.org/10.20344/amp.20721>

AUTHOR CONTRIBUTIONS

All authors contributed equally to this manuscript.

PROTECTION OF HUMANS AND ANIMALS

The authors declare that the procedures were followed according to the regulations established by the Clinical Research and Ethics Committee and to the Helsinki Declaration of the World Medical Association updated in 2013.

DATA CONFIDENTIALITY

The authors declare having followed the protocols in use at their working center regarding patients' data publication.

PATIENT CONSENT

Obtained.

COMPETING INTERESTS

The authors have declared that no competing interests exist.

FUNDING SOURCES

This research received no specific grant from any funding agency in the public, commercial, or not-for-profit sectors.

