

Images of Interest / Imagens de Interesse

Mazabraud Syndrome – Typical Radiological Findings of a Rare Disease

Síndrome de Mazabraud – Achados Radiológicos Típicos de uma Doença Rara

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Abstract

We present the case of a 52-year-old female patient with a known diagnosis of Mazabraud syndrome, involving the lower limbs and a history of total right hip replacement. She was referred to the Hospital das Clínicas in São Paulo, Brazil, after developing a non-painful swelling in the second left intermetacarpal space and diffuse hand oedema, with a one-year progression. As part of her evaluation at the institution, the patient underwent radiographic and MRI studies of the left hand, which confirmed the pathognomonic features of the known syndrome.

Mazabraud syndrome, a rare condition with fewer than 100 cases reported, is characterized by the association of fibrous dysplasia (typically polyostotic) and intramuscular myxomas. It is linked to genetic mutations and predominantly affects the lower limbs.

Keywords

Mazabraud syndrome; Intramuscular myxomas; Fibrous dysplasia.

Resumo

Apresentamos um caso de uma doente do sexo feminino, com 52 anos, com síndrome de Mazabraud conhecido e alterações que envolviam os membros inferiores tendo sido já submetida a prótese total da anca direita. Foi encaminhada para o Hospital das Clínicas de São Paulo (Brasil) após aparecimento de uma tumefação não dolorosa no 2º espaço intermetacárpico e edema difuso da mão, com um ano de evolução. Como parte da avaliação na instituição, a doente foi submetida a estudos radiográfico e de RM da mão esquerda, que confirmaram os achados patognomônicos do síndrome conhecido.

A associação entre displasia fibrosa, geralmente poliostótica e de surgimento mais precoce, com mixoma intramuscular, de aparecimento mais tardio, corresponde a uma entidade conhecida como Síndrome de Mazabraud. Trata-se de uma patologia rara, com menos de 100 casos descritos na literatura, associada e mutações genéticas e que afecta sobretudo os membros inferiores.

Palavras-chave

Síndrome de mazabraud; Mixomas intramusculares; Displasia fibrosa.

Case Description

We present the case of a 52-year-old female patient with a known diagnosis of Mazabraud syndrome, involving the right lower limb, previously treated with a total right hip replacement to correct a bone deformity. She was referred to the Hospital das Clínicas in São Paulo, Brazil, for evaluation of a painless swelling in the second left intermetacarpal space and diffuse hand oedema, with a one-year progression.

Initial radiographic studies (Fig. 1) revealed extensive intramedullary bone lesions with a radiolucent, ground-glass appearance, accompanied by endosteal scalloping and cortical thinning. The involvement was diffuse and multifocal, affecting the fingers, carpal bones, and distal radius and ulna, consistent with polyostotic fibrous dysplasia.^{1,2} Additionally, a soft tissue density was noted in the second intermetacarpal space, causing molding and remodeling of the radial border of the third metacarpal.

Subsequent MRI confirmed the presence of an intramuscular mass, hypointense on T1, hyperintense on T2 with heterogeneous post-contrast enhancement (Fig. 2), consistent with an intramuscular myxoma.¹



Figure 1 – Left hand radiograph (two projections) demonstrating diffuse and multifocal intramedullary bone lesions with a radiolucent, ground-glass appearance, consistent with polyostotic fibrous dysplasia. A soft tissue density is observed in the second intermetacarpal space, causing molding and remodeling of the radial border of the third metacarpal.

Discussion

The association of fibrous dysplasia with intramuscular myxomas defines Mazabraud syndrome, a rare condition with



Figure 2 – Axial and coronal T1-weighted (A, D), T2-weighted with fat suppression (B, E) and T1-weighted fat-suppressed post contrast (C, F) MRI images reveal a large intramuscular mass in the second intermetacarpal space, characterized by low T1 signal, high T2 signal and heterogeneous post-contrast enhancement, consistent with an intramuscular myxoma.

fewer than 100 cases reported in the literature.⁴ Recent studies have linked this syndrome to specific genetic mutations.^{1,2,3,4} Fibrous dysplasia results in fragile bones with pain, deformities, pathological fractures and functional impairment. It is predominantly polyostotic, most frequently involving the pelvis and femur. Radiographs characteristically reveal a radiolucent lesion with a “ground-glass” matrix and a sclerotic bone rim, while MRI typically demonstrates low T1, high T2 signals, and variable contrast enhancement.^{2,3} Although malignant transformation is exceedingly rare, the possibility of aggressive forms of fibrous dysplasia necessitates long-term follow-up.²

Intramuscular myxomas, another hallmark of Mazabraud syndrome, generally present as lesions with low intensity on T1-weighted images, high intensity on T2-weighted images and heterogeneous contrast enhancement on MRI.²

Imaging studies, particularly MRI, which is the gold standard, play a vital role in accurately diagnosing and monitoring the condition, ensuring early detection and timely management of clinical complications.⁴

Ethical Disclosures / Divulgações Éticas

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Confidencialidade dos dados: Os autores declaram ter seguido os protocolos do seu centro de trabalho acerca da publicação dos dados de doentes.

Protection of human and animal subjects: The authors declare that the procedures followed were in accordance with the regulations of the relevant clinical research ethics committee and with those of the Code of Ethics of the World Medical Association (Declaration of Helsinki).

Proteção de pessoas e animais: Os autores declaram que os procedimentos seguidos estavam de acordo com os regulamentos estabelecidos pelos

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