Lumps in the thigh

Mark S. Igra¹ • James D. Thomas²

Published online: 4 April 2016
© ISS 2016

Answer

Mazabraud Syndrome

Discussion

Mazabraud syndrome is an association between fibrous dysplasia and intramuscular myxomas, first described in 1926 by Henschen [1] and later by Mazabraud and Girard [2]. The syndrome has a mean presenting age of 46, with a range of 17–82. Roughly 70 % of cases are in women [3]. A review of 80 previous cases showed that 26 (33 %) had an association with McCune-Albright syndrome [4]. A recent case demonstrated the typical findings of this association: polyostotic fibrous dysplasia, multiple myxomas, brown-pigmented areas of the skin, and endocrine dysfunction [5].

Fibrous dysplasia is caused by a mutation to guanine nucleotide-binding protein, α -stimulating activity polypeptide 1 (GNAS1) [6], which encodes a protein involved in cell proliferation activity. The process is characterized by replacement of bone marrow by fibro-osseous tissue [7], may be mono- or polyostotic and is commonly unilateral.

Histologically, affected bones classically shows thin, irregular trabeculae and fibrotic marrow space, resembling Chinese lettering. It has an extremely varied radiological appearance on plain radiographs and CT, with ground-glass opacities, lucent foci or sclerotic areas. There may be bone deformity, expansion or endosteal scalloping. The polyostotic form of fibrous dysplasia predominates [8] and is most commonly seen in the femur and pelvis.

Myxomas are benign neoplasms that present in the 6th or 7th decade of life. In Mazabraud syndrome, they are usually diagnosed after or at the same time as fibrous dysplasia [9]. The myxomas are often multiple (70 % of cases) and non-contiguous with bone, although often found in close proximity with the area of fibrous dysplasia, as in the case presented here. Lesions have a predilection for the lower limbs. In Mazabraud syndrome, fibrous dysplasia is monostotic in 20 % of cases, and polyostotic in 80 % [9]. They are frequently asymptomatic, and patients may only present when the lesions are large enough to cause pain, restrict movement or cause vascular or neural impingement due to local mass effect. Our patient presented on noticing a large palpable mass in her vastus medialis muscle. A second smaller mass in vastus intermedius was not palpable.

Histopathologically, myxomas show bipolar and bland stellate fibroblasts separated by extracellular myxoid matrix. A diagnosis is strongly suggested if the lesions demonstrate typical myxoid MRI findings, including high water content, modest surrounding oedema and minimal surrounding fat atrophy. They are well-circumscribed low signal on T1-weighted images and high signal on T2-weighted images. In circumstances where these features are found in association with fibrous dysplasia, biopsy would not be required.

As there have been five reported cases of malignant change of fibrous dysplasia [10] and approximately 22 % of patients require surgery following pathological fracture [9], repeat

The case presentation can be found at doi: 10.1007/s00256-016-2385-8

✉ Mark S. Igra
dmmarkigra@gmail.com

¹ Sheffield Teaching Hospitals NHS Trust, Glossop Road, Sheffield S10 2JF, UK

² Radiology Department, Nottingham University Hospitals NHS Trust, B floor, Queen's Medical Centre Campus, Derby Road, Nottingham NG7 2UH, UK

imaging is advised if there are recurrent or new symptoms. Myxomas should be excised only if patients are symptomatic.

The combination of fibrous dysplasia and myxomas in Mazabraud syndrome is rare and likely to be under-diagnosed, and the risk of malignant transformation requires a high level of suspicion for the condition. Mazabraud syndrome has also been associated with uterine fibromas and adenofibromas, pagetoid skull, hamartomas and lipoma [5]. Mazabraud syndrome should be considered when patients present with any of these pathologies or associated conditions such as McCune-Albright syndrome.

Compliance with ethical standards

Conflict of interest The authors declare that they have no conflict of interest.

References

1. Henschen F. Fall von ostitis fibrosa mit multiplen tumoren in der umgebenden muskulatur. *Verh Dtsch Ges Pathol.* 1926;21:93–7.
2. Mazabraud A, Semat P, Roze R. A propos de l'association de fibromyxomes des tissus mous a la dysplasie fibreuse des os. *Presse Med.* 1967;75:2223–8.
3. McLaughlin A, Stalley P, Magee M, et al. Correlative imaging in an atypical case of Mazabraud syndrome. *Am J Roentgenol.* 2007;189:6.
4. Tagliafico A, Succio G, Martinoli C, Serafini G. Clinical overlap between Mazabraud and McCune–Albright syndromes. *J Ultrasound Med.* 2009;28:397–9.
5. Kitagawa Y, Ishihara Y, Hayashi M, Kim Y, Fujii N, Ito H. Mazabraud syndrome associated with McCune–Albright syndrome. *J Orthop Sci.* 2011;16:129–32.
6. Case DB, Chapman Jr CN, Freeman JK, Polga JP. Best cases from the AFIP: atypical presentation of polyostotic fibrous dysplasia with myxoma (Mazabraud syndrome). *Radiographics.* 2010;30(3):827–32.
7. Cohen Jr MM, Howell RE. Etiology of fibrous dysplasia and McCune–Albright syndrome. *Int J Oral Maxillofac Surg.* 1999;28:366–71.
8. Logel RJ. Recurrent intramuscular myxoma associated with Albright's syndrome. *J Bone Joint Surg Am.* 1976;58(4):565–8.
9. Zoccali C, Teori G, Prencipe U, Erba F. Mazabraud's syndrome: a new case and review of the literature. *Int Orthop (SICOT).* 2009;33:605–10.
10. Crawford EA, Brooks JS, Ogilvie CM. Osteosarcoma of the proximal part of the radius in Mazabraud syndrome: a case report. *J Bone Joint Surg Am.* 2009;91:955–60.