Case Report

Mazabraud syndrome

Anulekha Mary John, Kishore Kumar Behera, Thomas Mathai[1], Harshad Parmar[2], Thomas V. Paul

Departments of Endocrinology, Diabetes and Metabolism, [1]Orthopedics and [2]Pathology, Christian Medical College, Vellore, India

ABSTRACT

A 25 year old lady presented with pain and swelling of left thigh. On examination she was found to have tenderness of left femur with a separate soft tissue swelling within the thigh muscle. Further evaluation revealed expansile bony lesion on X-ray of left tibia and multiple hot spots on bone scan suggestive of fibrous dysplasia. The soft tissue swelling on excision and histopathological examination was found to be intramuscular myxoma. The combination of the above two, called Mazabraud syndrome is being reported.

Key words: Fibous dysplasia, intramuscular myxoma, mazabraud syndrome

Introduction

Mazabraud syndrome is a rare disorder of fibrous dysplasia associated with intramuscular myxoma. Both of these were identified at the same time in this particular patient.

Case Report

25 year old lady presented with swelling and pain of left thigh of seven years duration. This gradually increased in size without weight loss or proximal muscle weakness. She did not have difficulty in walking and her activities of daily living were not affected by the pain. Examination of the left thigh revealed bowing of the left femur and a soft tissue swelling not attached to the bone. The size of the swelling was 2 × 3 cm, mobile and mildly tender to palpation.

She had attained menarche at the age of 13 years and had normal menstrual cycles. She was married for the past one and a half years and had not conceived yet. Her body mass index was 25.1 kg/m^2 with height of 150 cm. She did not have any bony deformity or café au lait spots. On biochemical evaluation she had normal alkaline phosphatase, normal serum calcium, phosphorous and 24 hrs urine calcium, phosphorous and creatinine. She had a low level of 25 hydroxy vitamin D of 4.85 ng/ml (20.0-32.0 ng/ml) X-rays of pelvis and hip showed expansile bony swelling with cortical thinning of both tibia [Figure 1]. She underwent a bone scan which showed abnormal, irregularly increased tracer activity in left scapula, left tibia, left femur, pubis and left sacroiliac joints, L5 vertebra, left distal humerus and multiple ribs [Figure 2], suggestive of polyostotic fibrous dysplasia. MRI of the left thigh showed well defined intramuscular mass lesions [Figure 3]. These lesions were hypointense on T1 weighted image and hyperintense on T2W. They were partly exophytic and measured: 28 × 41 mm in the anteromedial aspect of right adductor longus, 16 × 23 mm in the right adductor magnus at the level of mid-thigh and 9 × 10 mm in the gluteus maximus A bone biopsy [Figure 4] from the left femur revealed fibrous dysplasia and a biopsy of the soft tissue mass from left thigh was confirmed as intramuscular myxoma [Figure 5].

Discussion

Polyostotic fibrous dysplasia in association with intramuscular myxomas is a rare condition. Only few case reports are mentioned in the literature and this is known as Mazabraud syndrome.[1] Both the lesions can occur in the same anatomical location as in our patient. Intramuscular myxomas associated with polyostotic fibrous dysplasia usually occur in multiple sites and present in adulthood while fibrous dysplasia occur at younger age.[1] Most of

Access this article online	
Quick Response Code:	Website: www.ijem.in
	DOI: 10.4103/2230-8210.113773

Corresponding Author: Dr. Thomas V. Paul, Department of Endocrinology, Diabetes and Metabolism, Christian Medical College, Vellore - 632 004, India. E-mail: thomasvpaul@yahoo.com

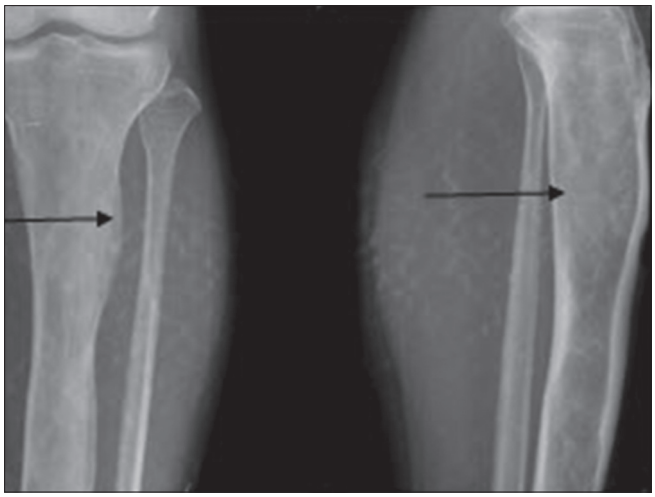

Figure 1: Anteroposterior radiograph of both tibia showing areas of mild expansile remodeling with endosteal scalloping and cortical thinning. There are regions of increased opacity of the bone showing ground-glass appearance characteristic of fibrous dysplasia

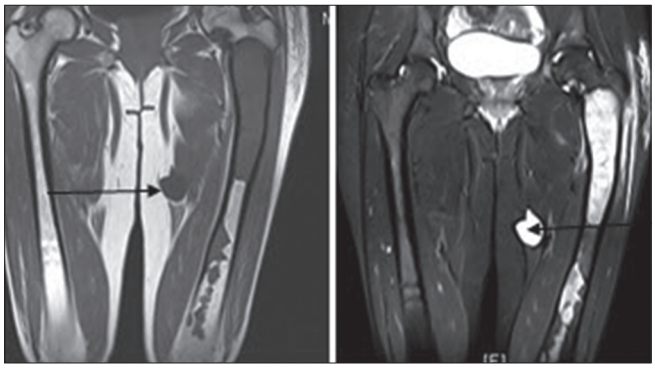

Figure 3: MRI of pelvis showing a well demarcated, intramuscular soft-tissue mass with low signal intensity (SI) on T1-weighted image and a high SI on T2-weighted image consistant with intramuscular myxoma

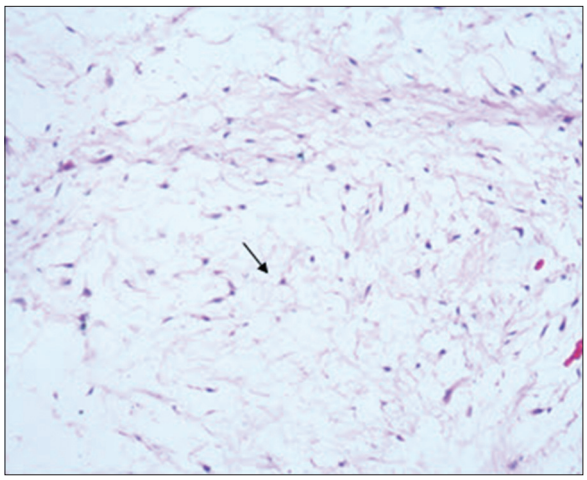

Figure 5: Biopsy of intramuscular myxoma showing loosely arranged haphazard fascicles of spindle to stellate shaped cells embedded in a hypovascular myxoid stroma (H&E stain at 10x magnification)

the patients usually present with minimal symptoms which can cause delay in diagnosing this condition. Both benign

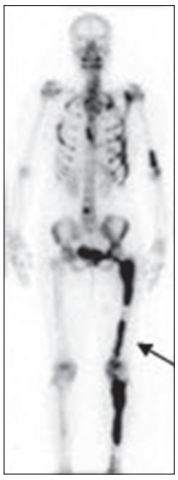

Figure 2: Technetium-99m methylene-diphosphonate bone scan revealed abnormal areas of radiotracer uptake involving left proximal femur, left humerus, left tibia multiple ribs and sternum

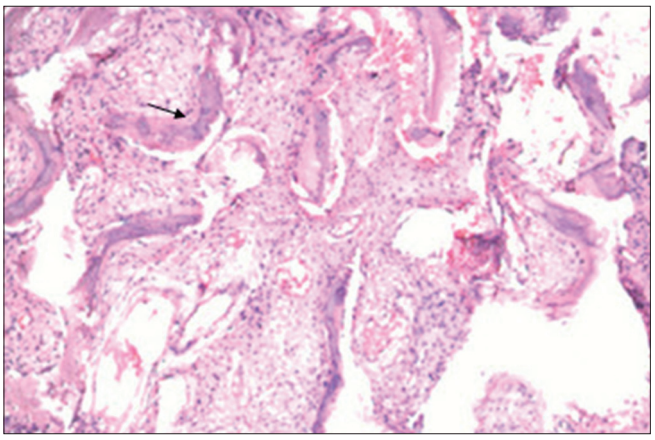

Figure 4: Bone biopsy showing irregular trabeculae of woven bone lacking osteoblastic rimming, set in a fibrous stroma. (Hematoxylin & Eosin stain, 10 x magnification)

and malignant myxoma can be confused with intramuscular myxoma which can only be confirmed by histopathology examination. These lesions are usually benign though local recurrence can occur if incompletely excised.

Though not common, malignant transformation of fibrous dysplasia can also occur as part of Mazabraud syndrome.[2] However, Mazabraud syndrome is so rare that only four cases of sarcomatous degeneration of fibrous dysplasia have been reported in the literature.

Myxomas in this syndrome are exclusively intramuscular and usually affect middle-aged women. Most common location is the thigh. Multiple intramuscular myxomas are usually associated with polyostotic fibrous dysplasia as in mazabruad syndrome. The treatment of intra-muscular myxomas is wide excision as local recurrence is reported. Sometimes it may enlarge to enormous size if not treated early. Late recurrence is also reported in the literature and hence long term follow up is warranted.[3]

Our patient underwent excision of the intra muscular myxoma and histopathology revealed a benign lesion. Though fibrous dysplasia could have been treated with bisphosphonates, it was deferred in this lady in view of her lack of bone pain, vitamin D deficiency and desire for fertility.

References

1. Kabukcuoglu F, Kabukcuoglu Y, Yilmaz B, Erdem Y, Evren I. Mazabraud's syndrome: Intramuscular myxoma associated with fibrous dysplasia. Pathol Oncol Res 2004;10:121-3.
2. Lopez-Ben R, Pitt MJ, Jaffe KA, Siegal GP. Osteosarcoma in a patient with McCune-Albright syndrome and Mazabraud's syndrome. Skeletal Radiol 1999;28:522-6.
3. Szendrói M, Rahóty P, Antal I, Kiss J. Fibrous dysplasia associated with intramuscular myxoma (Mazabraud's syndrome): A long-term follow-up of three cases. J Cancer Res Clin Oncol 1998;124:401-6.

Cite this article as: John AM, Behera KK, Mathai T, Parmar H, Paul TV. Mazabraud syndrome. Indian J Endocr Metab 2013;17:740-2.

Source of Support: Nil, **Conflict of Interest:** None declared.