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Case report

Mazabraud's syndrome: Report of its first incidence in the Middle East and a literature review

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HIGHLIGHTS

- This is the first case report of Mazabraud's Syndrome from Middle East.
- Providing a literature review of the disease and its presentation.
- Discussing GNAS1 gene involvement in fibrous dysplasia as well as other diseases.
- Documenting the management plan done for the patient and the result of the follow up.

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ABSTRACT

Introduction: Mazabraud's syndrome, a rare benign disease with indolent course, is best described as an association between soft tissue myxoma and fibrous dysplasia of the bones. In this report, we describe the first case of this syndrome from Saudi Arabia.

Case presentation: A 24-year-old male in overall good health status, presented with progressive left knee swelling over 6 years with no other associated symptoms. The swelling measured 5 cm in diameter, with smooth surface, and soft palpable texture. Radiological examination followed by histopathological examination of the excised mass confirmed our diagnosis of Mazabraud's syndrome. The patient was closely followed up with systematic examination with no recurrence.

Discussion: Fibrous dysplasia, soft tissue myxoma and multiple endocrinological diseases like McCune-Albright syndrome characterize Mazabraud's syndrome. Furthermore, fibrous dysplasia is found to be associated with GNA1S gene mutation. Many patients can have asymptomatic course of the disease but may present with pathological fractures, pain, and limitation of movement when the myxoma is near the joints or just simple cosmetically disturbing swelling like in our case.

Conclusion: Patients with such presentation need to be investigated thoroughly to rule out associated diseases and to evaluate the extent of such pathology. The improvement of radiological modalities can help in narrowing the differential diagnosis and following the patient to early detect the recurrence or any malignant transformation of the condition.

1. Introduction

Henschen first reported Mazabraud's Syndrome in 1926 [1]. The classic description of benign soft tissue myxomas associated with fibrous dysplasia of bone was later on detailed by Mazabraud et al. [2] To date, less than 100 cases of Mazabraud's Syndrome have been described in the literature [3,4] with more of female predominance [5].

Herein, and as to the best of our knowledge, we present the first case of this syndrome from Saudi Arabia and Middle East, with a brief review of the literature related to such presentation.

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2. Case presentation

A 24-year-old male patient who denies any past medical or surgical history with an overall general well being, presented to Plastic surgery outpatient clinic with a complaint of persistent swelling over the left knee that started 6 years prior to his presentation. The swelling was slowly progressive in size and not associated with pain. The patient denies any history of trauma to the site or history of other masses in the body. There was no history of discharges from the mass. It does not affect his gait or daily activity. Family history was unremarkable together with in-depth systematic review.

On physical examination, this was a young male patient with good body built. No skin lesions or obvious deformity were noticed. Chest, cardiovascular, and abdominal examination were within normal.

On local examination, a round swelling over the anterior aspect of the proximal tibia, 5 cm in diameter, No scars or punctum were seen and the overlying skin was intact. A smooth, non-tender, soft tissue mass was felt. It was not attached to the overlying skin, nor tethered to the underlying tibia. Left knee range of motion was full. Trans-illumination test together with Doppler signals were negative.

The patient was admitted to the hospital for further investigations and possible excisional biopsy, which was discussed with the patient. Routine labs were within normal limits.

In radiological assessment, X-ray of the left leg showed mild increased bone density, subtle irregular lucency and mild expansion with thickening of the cortices involving the proximal metaphysis and diaphysis of left tibia. No periosteal reaction was seen. There was a large antero-medial soft tissue swelling at the level of tibial tuberosity (Fig. 1). The team elected to further investigate the patient with MRI of the left knee, tibia and fibula that showed intramedullary abnormal signal changes involving the proximal tibial metaphysis and diaphysis. A lesion with well-defined borders and endosteal scalloping without cortical break through or periosteal reaction was found. The lesion had intermediate intensity on T2-weighted cuts and low signal intensity on T1-weighted images with patchy enhancement on the images post contrast. The most likely findings represent fibrous dysplasia. There were multiple small patchy areas within the tibial and femoral epiphysis, which showed patchy enhancement, likely representing polystotic fibrous dyplasia. There was a subcutaneous multi-loculated multi-septated cystic lesion seen within the most superior part of the anterior medial aspect of the left leg measuring 6.6 × 5.5 × 7.4 cm with pressure effect on the proximal tibia without direct invasion. It showed septation and capsular enhancement. It was either representing ganglion cyst or soft tissue myxoma. There was no evidence of joint pathology. The visualized muscle was grossly unremarkable (Fig. 2).

Based on the findings described above, Mazabraud's syndrome was considered as working diagnosis till definitive diagnosis can be achieved with excisional biopsy and histopathological assessment. Intra-operatively the mass was found to have dense attachment to

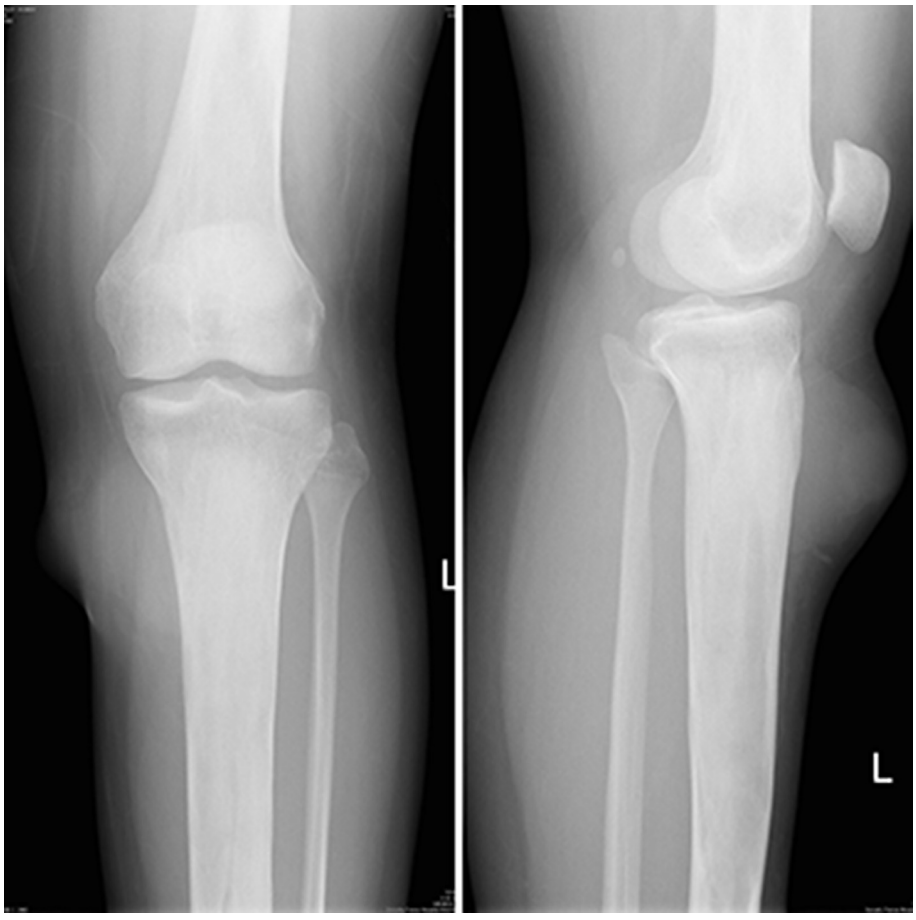

Fig. 1. X-ray of the left leg showed non-specific mild increased bone density, subtle irregular lucency and mild expansion with thickening of the cortices.