Mazabrauds syndrome an atypical presentation

Clinical History:

A 76 year old lady was referred with increasing paraspinal and left buttock pain over the past year. She had a previous liposarcoma excised from her left gluteal region 25 years ago. There was no history of systemic upset or recent weight loss. On examination, no masses were palpable.

Imaging Findings:

MRI was subsequently performed for further evaluation. This shows multiple osseous lesions associated with well-defined soft tissue masses with the largest one in the left gluteal region. The osseous lesions are heterogeneously intermediate on T1W (figure 1a) and heterogeneously high on STIR (figure 1b), consistent with fibrous dysplasia. The soft tissue masses are homogeneous with low signal on T1W (figure 2a) and high intensity on STIR images (figure 2b). The post-contrast images demonstrate heterogeneous central enhancement (figure 3b) when compared to the non-contrast T1W (figure 3a). Given the previous history of liposarcoma a biopsy was performed to exclude disease recurrence and the histology result confirmed that the lesions are myxomas.

Discussion:

Mazabrauds syndrome is a rare syndrome first described in 1967, characterised by the association of fibrous dysplasia, which can involve one bone (monostotic) or greater than one (polyostotic), and single or multiple benign intramuscular myxomas[1]. The aetiology of the syndrome is unknown, it has been proposed that the somatic mutations in the GNAS1 gene play a role[2, 3]. The gene has also been described in disorders such as McCune-Albright syndrome (triad of polyostotic fibrous dysplasia, café-au-lait spots and precocious puberty)[4, 5] and fibrous dysplasia. The intramuscular soft tissue myxomas component of Mazabrauds syndrome typically presents in the 6th decade and follow a benign course. These lesions are considered to be soft tissue hamartomas with tumour like growth[1, 6].

The lower limbs are classically affected, most commonly the right[4]. Patients often present with long standing painless soft tissue masses, a history of bone pain or recurrent fractures secondary to the bone dysplasia[7]. The associated soft tissue myxomas are located in close vicinity to the osseous component[1]. The syndrome more commonly affects females[6, 8].

Fibrous dysplasia is a congenital process, where there is a localised defect in osteoblastic differentiation and maturation. The normal bone is replaced with fibrous stoma and islands of immature bone which predominantly develop in children and young adults. Fibrous dysplasia has a reported 1-8% risk of malignant transformation into...
osteosarcoma[7, 9].

Typically, radiographic appearances of myxomas show a non-specific soft tissue mass. On MRI, the masses are well-defined and homogenous with low signal on T1 and high signal on T2 weighted images as in our case. The post contrast images usually show heterogeneous central enhancement and also rim enhancement of the pseudo capsule [10]. In our case there was heterogeneous central enhancement but no convincing enhancement of the pseudocapsule. The soft tissue masses are closely associated with hyperintense osseous lesions on T2 weighted images, which are the regions of fibrous dysplasia.

Mazabrauds syndrome is generally managed conservatively, however in some cases the myxomas can be resected if they are causing symptoms but there is a high risk of recurrence [5, 10].

In conclusion Mazabraud syndrome should be considered early in patients presenting with fibrous dysplasia and soft tissue masses, especially if the soft tissue lesion is myxoid in nature, as it is generally managed conservatively.

Written informed patient consent for publication has been obtained

**Differential Diagnosis List:** Mazabrauds syndrome, Intramuscular myxoma, myxofibrosarcoma

**Final Diagnosis:** Mazabrauds syndrome

**References:**


Figure 1

Description: T1W images showing two left sided osseous lesions within the left femur and ilium, which have low signal on T1W Origin: Nottingham University Hospitals
Description: STIR images showing the two left sided osseous lesions within the left femur and ilium, have high signal on STIR. Origin: Nottingham University Hospitals
Description: T1W image showing two left sided soft tissue lesions with one seen in the gluteal region and the other one in the posterior femoral compartment with low signal on T1. Origin: Nottingham University Hospitals
Description: STIR image showing the left gluteal and proximal femoral soft tissue lesions which demonstrates high signal on STIR. The signal characteristics appear myxoid in nature. Origin: Nottingham University Hospitals
Figure 3

Description: T1W image shows homogeneously low signal lesion within the left gluteal lesion. Origin: Nottingham University Hospitals
Description: Post-contrast T1W(right) image showing heterogeneous central enhancement. Origin: Nottingham University Hospitals