Case Report

An atypical monomelic presentation of Mazabraud syndrome

Jun Wan, Hong-Bo He, Qian-De Liao, Can Zhang

Abstract

Mazabraud syndrome is a rare condition characterized by a combination of fibrous dysplasia and intramuscular myxomas. In Mazabraud syndrome, the distribution of fibrous dysplasia is mostly polyomelic and frequently located in the femur, with myxomas adjacent to the fibrous dysplasia lesion of bone (mostly in the quadriceps muscle). However, when presented as atypical clinical features, patients of Mazabraud syndrome is either misdiagnosed or difficult to diagnose. We report an atypical monomelic case of Mazabraud syndrome in the right upper arm and discuss the difficulties in making an accurate diagnosis.

Key words: Mazabraud syndrome, fibrous dysplasia, myxomas, monomelic, upper limb
MeSH terms: Fibrous dysplasia of bone, muscle, skeletal, upper extremity

Introduction

Mazabraud syndrome is rare benign disorder defined as combination of monostotic or polyostotic fibrous dysplasia with single or multiple intramuscular myxomas. Fibrous dysplasia developed in bone is a benign and fibro osseous lesion, and myxomas presented as benign mesenchymal tumors.[1] The first case was reported by Mazabraud,[2,3] which was finally named after his name. Although there is no evidence of continuity between the fibrous dysplasia and myxomas, the myxoma is generally located in the vicinity of the affected bones.[4] The onset of fibrous dysplasia often occurs at younger age, which predates the appearance of myxoma mostly presented in adulthood. The disease has a predilection of women than men and the etiology is still unknown. Some study by genetic analyses has shown certain point mutations in the GNAS-1 gene, which regulates the cellular monophosphate level in fibrous dysplasia and intramuscular myxomas.[5]

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Patients of Mazabraud syndrome are often asymptomatic. In general, fibrous dysplasia in Mazabraud syndrome was mostly polymelic which predominantly affect the femurs, tibias and pelvis. The most common location of myxoma is the thighs (quadriceps).[6-8] However, when presented as atypical clinical features, patients of Mazabraud syndrome are either misdiagnosed or difficult to diagnose. We report an atypical monomelic case of Mazabraud syndrome in the right upper arm and share our experiences in approaching the diagnosis.

Case Report

A 44 year old female patient, cotton spinner by occupation, presented with pain and a hard mass in the right upper arm which had gradually increased in size in the past 2 months. Physical examination revealed a painless oval soft-tissue mass within the biceps muscle of the right upper arm. There was no upper limb length discrepancy, cafe au lait spot or any endocrine problem. A plain x-ray of the right humerus showed a typical "ground-glass-like" lesion without obvious soft tissue swelling and periosteal reaction. However, the bone cortex seemed to have defects near the fibrous dysplasia of the proximal humerus [Figure 1a]. Computed tomography (CT) examination showed multiloculated radiolucent endosteal scalloping involving the full length of the right humeral shaft, combined with a solitary hypo-dense soft-tissue mass. It seemed that some continuity did exist between fibrous dysplasia of the proximal humerus with soft-tissue mass [Figure 1b]. Clinicoradiological diagnosis was suspected soft-tissue sarcoma secondary to fibrous dysplasia of the humerus. Radiotherapy and chemotherapy were suggested along with wide excision surrounding the mass or amputation of the right upper limb and patient was transferred to this institute.

In order to get more clues for further diagnosis, magnetic resonance imaging (MRI) examination of the right upper arm was performed [Figure 2a and b]. The results showed that the mass appeared a defined oval intramuscular (biceps muscle) type which was low signal intensity on T1-weighted images (T1WI), while homogeneous fluid-like high signal intensity on T2-weighted images (T2WI). Changes in intensity of the humerus were consistent with fibrous dysplasia. However, the bone cortex of the humerus was intact demonstrating that continuity with bone and soft tissue lesions was not apparent [Figure 2c and d]. These findings did not correlate with the initial diagnosis of previous hospital but implied some lesion like ganglion cyst (composed of gelatinous mucoid).

The radiological findings made the diagnosis more confusing. A further 99mTc-methylene diphosphonate (MDP) whole-body bone scan was performed and the result revealed no concentration at the site of the mass in the right biceps, but increased uptake in the right humerus. These changes were interpreted as monomelic fibrous dysplasia. Due to the difficulty of making diagnosis, biopsy of both soft-tissue mass (total excision) and adjacent humerus (local curettage and allografting) was performed. The histological findings were described as intramuscular myxoma of soft-tissue mass and fibrous dysplasia of the humerus lesions, both without evidence of sarcomatous degeneration [Figure 3a-c]. A final diagnosis of Mazabraud syndrome (one atypical monomelic type) was established by the histological results. No recurrence of myxoma and sarcomatous degeneration of fibrous dysplasia were observed in 3 years followup.

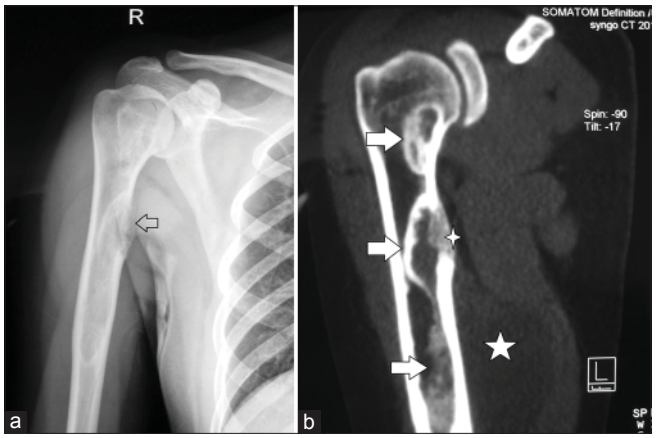

Figure 1: (a) Anteroposterior radiograph of the right humerus showing a diaphyseal lesion with a diffuse "ground-glass" appearance. A bony cortical lesion is also seen arrow (b) Coronal imaging of computed tomography of the humerus showing endosteal scalloping (white arrow) with a solitary hypodense intramuscular mass in the softtissue (white pentagram). Some continuity did exist between fibrous dysplasia of proximal humerus with soft-tissue mass (white star)

Discussion

The etiology of Mazabraud syndrome still remains unknown.[6,8,9] Considering the peculiar characteristic of myxomas tend to arise close to the affected segments of fibrous dysplasia, some studies regarded them as a reactive phenomenon due to local mechanical stress factor.[8] Actually, myxomas appear to be similar in histology to ganglion cysts which result from the regional stressed articulations. We believe that the regional mechanical stress factor played an important role in the development of myxomas near the site of fibrous dysplasia as well. We infer the myxoma occurred as a reaction to resist the stress.

In order to make proper diagnosis of Mazabraud syndrome, MRI was better recommended rather than plain X-ray and CT. The extent and association of fibrous dysplasia and myxoma can be more accurately defined in MRI.[10,11] Plain X-ray and CT examination would confuse to make an almost catastrophic misdiagnosis as in the first hospital. Further MRI examination in our hospital revealed the lesions in the right biceps have no contact with the adjacent humerus affected by fibrous dysplasia, which presented as low signal intensity

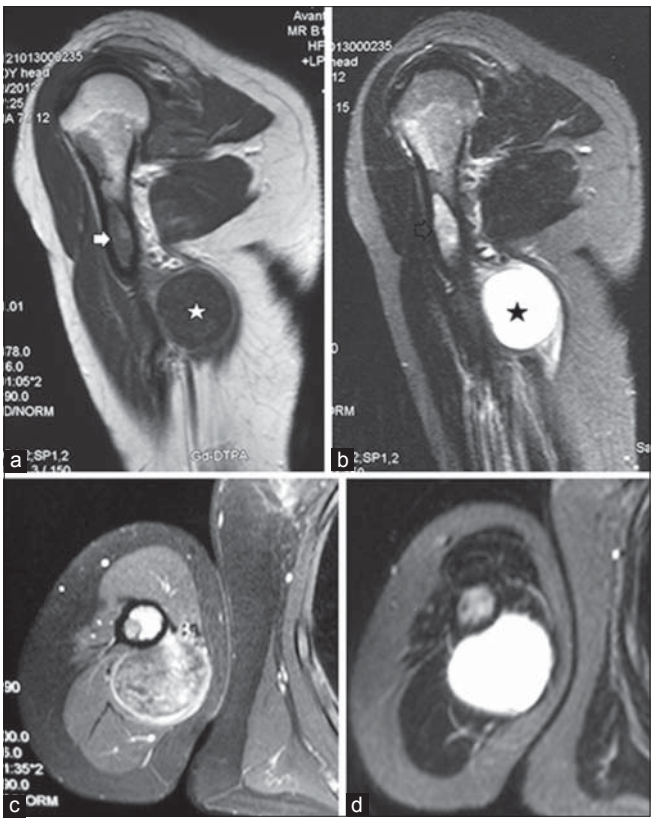

Figure 2: (a) T1-weighted MRI showing a low intensity softtissue mass and (b) a high intensity one of the right upper arm in T2-weighted images. Changes in intensity of the humeral shaft were thought consistent with fibrous dysplasia (white arrow). (c and d) The bone cortex of the humerus was intact which indicated continuity with bone and softtissue lesions were not apparent

Figure 3: (a) Gross specimen of the intramuscular mass showing a characteristic "gelatinous-mucoid-like" appearance. (b) Histological examination of the soft tissue mass showing that spindle cells are loosely arranged within an abundant loosely myxoid matrix. (c) Histological examination of the bone lesion revealed immature woven bone trabeculae laid in a proliferated fibrous connective tissue without osteoblastic lining (×200)

on T1WI and high signal intensity on T2WI. All those presentation implied that the lesion was not a "sarcoma-like" mass initially interpreted by CT,[12,13] but a "ganglion-cyst-like" mass. Whole body bone scan may demonstrate confluent asymmetric areas of high uptake, which can help to find out asymptomatic lesion of fibrous dysplasia.

To our knowledge, our case of Mazabraud syndrome demonstrated several atypical clinical features not noted before. Firstly the patient presented with solitary myxomas in the right triceps muscle region, rather than the thigh or pelvis, as reported in most cases. Secondly fibrous dysplasia occurred monomelic in the right upper arm (humerus) not multiple or in the lower limbs as in most cases. Lastly and most importantly, the presenting myxoma and all sites of fibrous dysplasia were only in a monomelic distribution in the right upper arm. The unique phenomena make it hard to differentiate from soft-tissue sarcoma arising from the fibrous dysplasia of adjacent bone.

Multidisciplinary approaches were recommended for diagnosis such as MRI,[11] whole-body bone scan examination and biopsy. Moreover, one thing should be mentioned that a greater risk of malignancy of fibrous dysplasia is present for patients with Mazabraud syndrome.[14,15] The long term followup is mandatory to find out the possibility of sarcomatous degeneration of fibrous dysplasia in patients of Mazabraud syndrome.

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