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Case Report

A Rare Case Of Maffucci's Syndrome. From The Diagnosis To The Management.

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Abstract

Multiple chondromas are infrequent, prevails in males, the most affected bones are the small tubular bones of the hand and foot, and in rare cases there are diffuse chondrodysplasia, known as Ollier's disease. We report a rare case of Maffucci's syndrome in a 47 year old woman with post-traumatic pains to fourth and fifth finger of her right hand.

Radiographic examination revealed pathological fracture of the proximal phalanx of the fourth and fifth fingers of her right hand associated with osteolytic lesions and multiple calcifications as commas, referring to lobulated cartilage masses deforming all phalanges of her fingers of right hand and phleboliths calcified inscribed in multiple cutaneous hemangiomas.

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The case report descirbe the importance of medical history and examination suggest that, the diagnosis of this rare anatomical-clinical entity with additional diagnostic (PET / CT) is important to exclude high risk of malignant transformation and to identify additional pathological sites.

Keywords: Maffucci's syndrome, enchondromatosis, multiple hemangiomas, pathological fractures.

Introduction

Maffucci's syndrome is a rare mesodermal dysplasia, not hereditary, characterized by multiple enchondromas and hemangiomas, especially in the extremities [(1)].

The enchondromas can be very exentesive, and generally grow near the accretion disk cartilaginous and can result from dysregulated proliferation and differentiation of chondrocytes during physiological endochondral ossification [(2)]. Radiographic pathognomonic sign is the presence of linear radiotransparency that from the metaphysis extend along the diaphysis of the long bones. 30% of this patients have phatological fractures, and a half of this fractures have late or missed consolidation. Complications include spontaneous fractures, skeletal deformities and the possibility of malignant transformation. Transformation in secondary sarcoma, most commonly chondrosarcoma, is frequent and probably ranges 20-30% in Ollier's disease and certainly higher in Maffucci (likely >50%) and may occur at every age, expecially in adult patient [(1)]. Three types of vascular lesions are associated with Maffucci's syndrome: cavernous hemangiomas, phlebectasias and lymphangiectasia-lymphangiomas. Disease may be associated with benign or malignant tumors, such as pituitary adenoma and parathyroid, astrocytoma, breast, ovarian cancer and CNS tumors [(3)].

The surgical treatment is aimed to relieve symptoms rather than excise chondromas. Prognosis is burdened by the incidence of malignant change. This disease usually affects only one side of the body. Bone and vascular lesions develop at birth or in early childhood and may be progressive[(3-4)]. The average age at diagnosis is 12 years and the diagnosis is based on clinics. The surgery is not required in the absence of complications, instead of the symptomatic lesions can be treated with curettage and bone graft[(5)].

A recent compares between immediately treatment and deferred treatment has show more complications for the immediately treatment (67% vesus 10%). Fixation is not required for the shot bone, but can be useful in case of femur and other long bones of upper and lower limbs fractures. Anyway, the treatment should be personalized with an appropriate pre-operative planning in all cases[(5)] .

Case report

A french female, 47 year old presents in our department of the Hospital Santa Maria della Misericordia, Perugia with pain at the base of the fourth and fifth fingers of the right hand after a trauma.

Patient reported to be affected, since she was 5 years old, by a deformity of the last two fingers of her right hand, associated with ankylosis of the interphalangeal joints, both distal and proximal. At the clinical examination she has the entire right arm covered with multiple hemangiomas skin and swelling of the fourth and fifth fingers of her right hand. The radiographic examination, performed in back-projection handheld (Figure 1)

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and oblique projection(Figure 2) showed the pathological fracture of the proximal phalanx of the fingers, affected by this disease. In addition, there were osteolytic lesions associated with multiple punctate calcifications as commas, referring to the lobulated cartilage masses enough to deform greatly all the two phalanges examined. Finally, authors noted the presence of calcified phleboliths in multiple cutaneous hemangiomas. In the laboratory test there were only three component of blood test abnormal: the VES was higher 71, the Calcium was 9,1; alkaline phosphatase 83 IU. According to the clinical signs and medical history and radiological findings it was hypothesized diagnosis of Maffucci's syndrome. We performed radiological comparison between the right and the left hand and also in the left hand there are a little lesion; in fact we could see at the diaphyses of the fifth metacarpus a single oval osteolytic lesion with peripheral sclerotic margin and with a maximum diameter of about 7 mm to report to enchondroma (Figure 3-4 arrow blue).

We also studied the old radiographic imaging that the patient had done three years ago for a rib fracture, and we showed a similar lesion on the right scapula and millimetre multiple osteolytic lesions intermingled to enchondromatosis area, despite of she had not clinical signs of pain and / or functional impairment of the shoulder . On the scapula we could see dysmorphic area on the medial edge, the margin lower side, the upper and the coracoid process; furthermore, it was reported a single oval osteolytic lesion with sclerotic margin and a maximum diameter of 11 mm to the neck of the scapula, compatible to enchondromatosis. (Figure 5-6).

After this discoveries of the enchondromatosis injuries founded at radiographic imaging, we proposed to the patient to undergo investigation Pet-CT with F-18FDG. This examination confirmed that the locations identified by radiography and excluded at the same time further injuries in other body areas and this lesion are only benign. (Figure 7-8) The uptake of the radionuclide was modest level of 4 and 5 finger right, slightly picked up to level 5 metacarpal on the left and the right shoulder blade.

In regard of this kind of multiple benign lesion, we decided that our treatment is conservative, so periodic follow up every three month with radiographic imaging to evaluate the enhancement of this enchondromas.

Actually at the first follow up, the patient have not pain or functional impairment of the shoulder, she soffers only for swelling of the finger and for the ankylosis but she has not got any kind of fracture.

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CONFLICT OF INTEREST STATEMENT: All authors must disclose any financial and personal relationships with other people or organisations that could inappropriately influence (bias) their work. Examples of potential conflicts of interest include employment, consultancies, stock ownership, honoraria, paid expert testimony, patent applications/registrations, and grants or other funding.

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Figure 1: Right Hand's XR in AP shows two big enchondroma that involve the whole last two fingers and whole metacarpal bone.



Figure 2: Right Hand's XR in Oblique shows two big enchondroma that involve the whole last two fingers and whole metacarpal bone.

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Figure 3: Left Hand's XR in AP shows the 7mm's enchondroma (arrow).



Figure 4: Left Hand's XR in Oblique shows the 7mm's enchondroma (arrow).



Figure 5: PA XR shows a similar lesion on the right scapula and millimetre multiple osteolytic lesions intermingled to enchondromatosis area.



Figure 6: AP XR of the right shoulder shows a single oval osteolytic lesion with sclerotic margin and a maximum diameter of 11 mm to the neck of the scapula, compatible to enchondromatosis

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Figure 7: The Fused Sagittal Pet-CT exluded other osteolytic skeletal area



Figure 8: The Sagittal Pet-CT exluded other osteolytic skeletal area.

Discussion

Maffucci syndrome is a very rare condition; there are just about 200 cases published in the literature [(6)]. About 25% of patients come into contact with signs and symptoms of Maffucci's syndrome at birth, 45% before the age of 6 and 78%, as our patient, symptoms occur before puberty. It is classified as a mesodermal dysplasia that manifests itself as a combination of multiple hemangiomas enchondromatosis[(7)].

The enchondromatosis is a disease characterized by the presence of multiple enchondromas, typically in correspondence of the metaphysis and the diaphysis of bone. The clinical manifestations of multiple enchondromas such swellings gnarled fingers (as in our case) or obvious differences in the length of the forearms or legs, generally are found in childhood and adolescence. This pathology prefers a side of the body. The syndrome has not familiar or genetic transmission [(8)]. Lewis and Ketcham describe the anatomical distribution of the bone lesions in their study. In their analysis of 98 cases, they found that the hand is often involved (88%), followed by the foot (and the leg (61-59%), the femur (53%), the humerus and forearm (41%), the pelvis (21%) and by the vertebrae (10%).[(7)] Recent studies indicate that the presence of multiple enchondromas at the hand of the patient is the most common location of the people affected by this syndrome, but the scapula's location is very rare. Both the bone injuries of the hand and the scapula associated with soft tissue hemangiomas show a characteristic presentation : the preference for one side, in this case the right

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[8]. The most frequent malignant tumor that could occour is chondrosarcoma , but they have described other cancers including angiosarcoma and fibrosarcoma.[7] In a series of studies carried out on 105 patients with the Maffucci's syndrome, based on the number and type of cancer discoveries, there were 24 malignant tumors (23% of cases), including 16 chondrosarcoma, 2 gliomas, 2 ovarian tumors mesenchymal and individual cases of hemangiosarcoma, lymphangiosarcoma, pancreatic cancer and fibrosarcoma [7]. In addition, Maffucci's syndrome patients have a high risk of malignant transformation of skeletal injuries and soft tissue after 40 years. In our case , the radiographic study of the fourth and fifth fingers of her right hand, and the right scapula has showed the presence of radiological findings characterized by osteolytic lesions associated with multiple punctate calcifications and a comma, to report to the masses cartilage lobulated enough to deform the bone structure; were also featured phleboliths calcified inscribed in multiple hemangiomas of the soft tissue of the upper right corner. This radiographic confirm this very rare syndrome. According to Muthusami et al. these multiple bone lesions, in the first hypothesis related to the Maffucci syndrome, should be differentiated from other disorders such as syndrome Ollier, Fibrous dysplasia Polyostotic, Hereditary Multiple Exostoses, Paget's disease and bone metastases [9-10]. In the study of bone lesions of this disease, it's very important to evaluate: the absence of hereditary genetic transmission or family nature; the young age and the seats of onset; the laboratory tests negative, only the levels of alkaline phosphatase and calcium are modified; the good health condition and finally the radiographic findings characterized by multiple radiolucent bone lesions -with central calcifications - that can erode cortical bone structure and deform it. Clinical and radiographic findings of soft tissue hemangiomas of the right arm, characterized by phleboliths calcified, allowed to reach definitive diagnosis. The case report emphasizes the importance of clinical-radiological history and medical examination to diagnose this rare anatomical-clinical entity and the PET / CT scan in order to exclude the high risk of malignant transformation of the lesions and to identify additional pathological sites.

References

1. Verdegaal SHM,et al.: Incidence, predictive factors, and prognosis of chondrosarcoma in patients with Ollier disease and Maffucci syndrome: an international multicenter study of 161 patients. Oncologist 2011, 16 (12): 1771-1779.

2. Amary MF, et al.: Ollier disease and Maffucci syndrome are caused by somatic mosaic mutations of IDH1 and IDH2 . Nat Genet 2011, 43 (12): 1262-1265.

3. Jermann M, Eid K, T Pfammatter, Stahel R: Maffucci's syndrome. Circulation 2001, 104 (14): 1693.

4. Gao H, Wang B, Zhang X, Liu F, Lu Y: Maffucci syndrome with unilateral limb: a case report and review of the literature. Chin J Cancer Res, 2013, 25 (2): 254-258.

5. Pannier S, Legeai Mallet-L: Hereditary multiple exostoses and enchondromatosis. Best Pract Res Clin Rheumatol 2008, 22 (1): 45-54.

6. Mustapha Amyere et wings: Common Somatic Alterations in Maffucci Syndrome Identified by Molecular Karyotyping, Molecular Syndromology, published online on 08.26.2014.

7. Lewis RJ, Ketcham AS. Maffucci's syndrome: functional and neoplastic significance.Case report and review of the literature. J Bone Joint Surg Am. 1973;55:1465–79.

8. Albregts AE, Rapini RP (1995) Malignancy in Maffucci's syndrome. Dermatol Clin 13:73-78

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9. Adam Greenspan. Imaging in orthopedics. Ed. IV Rome, Italy. International editions 2007. 614-619,630-641,646-648, 746-757, 845-861.

10. Muthusamy S, Conway SA, Temple HT: Five polyostotic conditions That general orthopedic surgeons Should Recognize (or Should not miss). Orthop Clin North Am. 2014 Jul; 45 (3): 417-29. doi: 10.1016 / j.ocl.2014.04.004.

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