A Lesson To Learn : A Rare Case Of Maffucci's Syndrome

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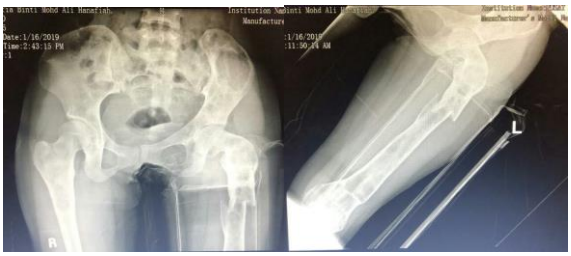
INTRODUCTION:

Maffucci's syndrome is a very rare mesodermal dysplasia, not hereditary, characterized by multiple enchondromas and hemangiomas especially in the extremities. The enchondromas can be very extensive, and generally grow near the accretion disk cartilaginous and can result from dysregulated proliferation and differentiation of chondrocytes during physiological endochondral ossification. Radiographic pathognomonic sign is the presence of linear radiotransparency from the metaphysis extend along the diaphysis of the long bones. 30% of this patients have patological fractures, and a half of this fractures have late or missed consolidation. Complications include spontaneous fractures, skeletal deformities and the possibility of malignant transformation. We report a rare case of spontaneous fracture in a patient with Maffucci syndrome as a learning purpose.

REPORT :

A twenty five years old lady with maffucci's syndrome presented with sudden onset of left hip pain while walking. Denied any history of significance trauma or fall. Upon physical examination noted deformity with bony tenderness over proximal left thigh. Radiograph findings showed irregularly shaped over cortical bone. Radiolucent areas were found with no stippled calcification within bilateral femur. Noted fracture over proximal third of left femur.



CONCLUSION:

Maffucci syndrome is a very rare condition. There are just about 200 cases published in the literature. Treatment for Maffucci syndrome should be aimed at early detection of malignant transformation as well as symptom relief. Surgery is indicated only in the case with complications, such as pathological fractures, growth defects and malignant transformation. The goal of surgery is to remove the tumor mass and make histological diagnosis.

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. 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