CASE REPORT

THE MAFFUCCI SYNDROME: A RARE CASE REPORT

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ABSTRACT

The Maffucci syndrome is a rare genetic disorder that affects both males and females. It is characterized by a benign tumor of the cartilage (enchondroma), bone deformities, and dark, irregularly shaped hemangiomas. It was first reported by Angelo Maffucci in 1881 -after a forty-year old woman died from complication following amputation of an arm. We are presenting Maffucci syndrome in a 15-year old Ethiopian female. To our knowledge so far, there is no such documented report from Ethiopia.

INTRODUCTION

The Maffucci syndrome is characterized by enchondromas, resulting in bone deformities and soft tissue hemangiomas. The chondromatous lesion and, less commonly, the hemangiomas can undergo malignant transformation. The risk for sarcomatous degeneration of enchondromas, hemangiomas, or lymphangiomas is 15-30%.Superficial and deep venous malformations frequently protrude as soft nodules or tumors usually on the distal extremities, but they can also appear elsewhere. The cause of the syndrome is unclear with no familial pattern of inheritance. It appears sporadically with no sex or race predilection. Patients are normal at birth and the syndrome manifests during childhood and puberty. The enchondromas affect the extremities; their distribution is asymmetrical leading to disfigurations. Pathological fractures which arise in affected metaphysis and diaphsis of the long bones are commonly encountered (In 26%).

The disease appears to develop around the age of 4 to 5 years in 25 % of the cases. In 45% of the cases symptoms start before the age of 6; and in 78% of the cases symptoms develop before puberty (1, 2, 3, 4).

The main aims of the management of the Maffucci syndrome involves early detection of malignancy and relief of symptoms. Asymptomatic patients need no treatment, but it is recommended that it have close follow-up to evaluate for any changes in the skin and bone lesions. Treatment options in symptomatic patients includes, Cosmetic surgery, fixation of pathological fracture, correction of deformity, and treatment of haemangioma Radical excision or amputation may be mandatory in cases of malignant transformation. The periodical evaluating team comprises of a radiologist, an orthopedist, and a dermatologist.

CASE REPORT

A 15-year old girl came to the Out Patient Department of 'Tikur Anbessa' specialized teaching hospital due to gross progressive deformity and multiple swellings of her left hand over the last eight years. The deformity and the swellings were associated with mild pain. She was X-rayed after history, and physical examination and laboratory investigations were done.

1. ***Physical findings:*** A gross deformity of the left-hand with non-tender, firm to hard multiple swellings was seen (9 in number). The largest soft tissue swelling is purplish and easily compressible. It was found at the lateral aspect of DIP of the ring finger.
2. ***Lab findings:*** Complete blood count, ESR, and serum calcium levels were normal.
3. ***Radiographic findings:*** Please note from picture-1:

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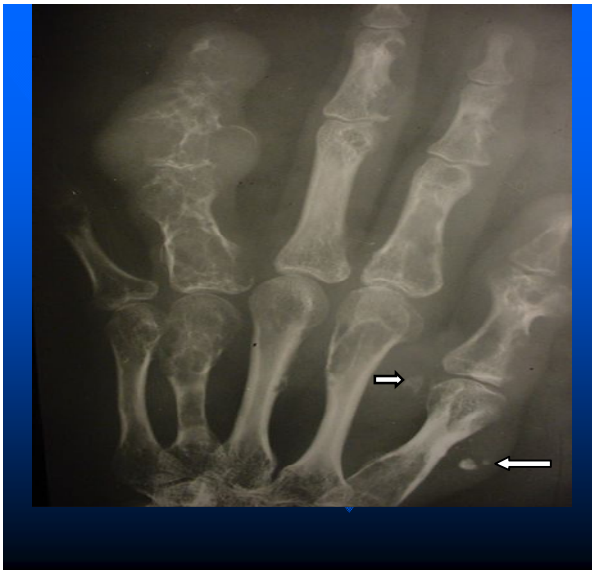
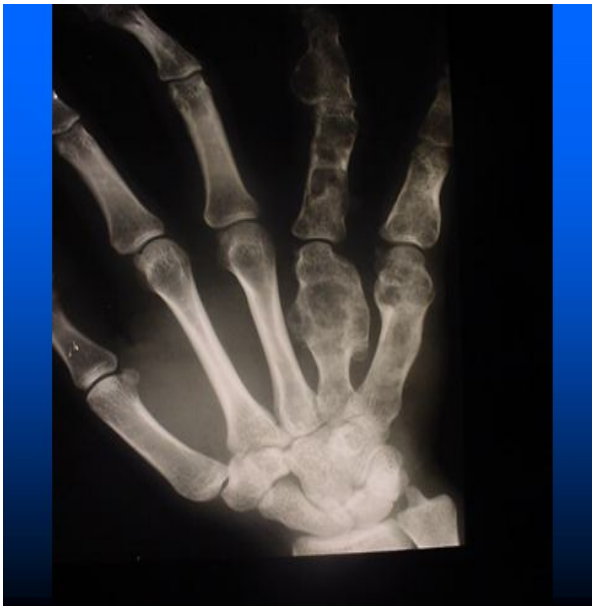
A. Multiple medullary lesions nearly in all metacarpals and phalanges.
B. Eccentric soft tissue swelling with adjacent cortical erosion of the fingers. The proximal phalynx of little finger is totally eaten-up.
C. Soft tissue swellings with phleboliths (*vascular calcifications of haemangioma*) on medial and lateral aspects of the thumb metacarpal bone (Shown by the arrows)



Picture 1. PA plain X-ray of a 15 year old girl presented to "Tikur Anbessa" Hospital with Maffucci syndrome.

Histopathological examination: Fragments of grey bony, hard specimen "lobules of different sizes separated by small septa of fibrous tissue, uniform cartilage cells with dark round nuclei & some binucleate nuclei are seen" with a thin layer of lamellar bone surrounding the cartilage nodules. Blood vessels are surrounded by osteoid with chondrocytes without atypia inside hyaline cartilage. The nuclei are small, round and pyknotic. The cellularity varies between lesions and within the same lesion. The cartilage lesions are very cellular and may have a large number of binucleated or atypical chondrocytes. The cellular atypia is far greater than what is seen in ordinary/simple enchondroma; rather it is common with the Mafucci syndrome.

Pathologists comment: Each potential enchondroma needs to be evaluated for cellularity, nuclear atypia, double nucleated chondrocytes, and mitotic activity in a viable area without calcifications to distinguish it from low-grade chondrosarcoma. The pathologic diagnosis is so difficult that it always needs to be made in conjunction with the radiologist and the orthopedic surgeon.



Picture 2. Ollier's disease (for comparison)
* Note that there are no calcifications.

DISCUSSION

The Maffucci syndrome is a rare, nonhereditary disease characterized by multiple enchondromas combined with hemangiomas and less commonly lymphangiomas (1,2). It is sometimes confused with Ollier's disease that consists of multiple enchondroma without hemangiomas (1,2). The dyschondroplasia in Maffucci syndrome is of unknown origin. There is a derangement of cartilaginous growth which results in the migration of cartilaginous rests from the epiphyseal plate into the metaphseal regions. Proliferation of these bits of cartilaginous tissue leads to the development of intraosseous chondroma which is usually located in the metaphysis (3,4). Intracortical enchondromas have been described by some authors (**5**).

The skeletal and vascular lesions in extremities are usually asymmetrically distributed with unilateral changes seen in about 50% of patients (2,**6**).The hemangiomas in Maffucci syndrome are mostly located in subcutaneous soft tissue and appear as blue subcutaneous nodules which can be emptied by means of manual compression. The skeletal lesions show a predilection for the involvement of tubular bones. The most frequent sites of enchondromas are the metacarpals and the phalanges of the hands and less commonly the feet (2).

Enchondromas can manifest as a pain less swelling of a digit or as pathological fractures. In this rare disease radiographs, particularly of the hands and feet, are often pathognomonic (1,2). The radiolucent skeletal lesions are well demarcated and show an expansile remodeling of the affected bone with a predominant thinning of the cortex and endosteal scalloping. Deformities of the extremities may be caused by an interrupted skeletal growth during development. In the soft tissues, phleboliths- which are typical of cavernous hemangiomas- and soft tissue calcification may be seen (3).

The risk of the Maffucci syndrome is a malignant transformation of the bone lesion, commonly to chondrosarcoma. The review of literature shows a discrepancy in the rate of malignant transformation of the skeletal lesion of the syndrome. In 1973, Lewis and Ketcham (**7**) reported that a sarcomatous transformation of an enchondroma is 15.2%. Other authors reported a risk of malignant trasformation varying between 30-56% (**8,9**). The risk of malignant transformation of skeletal lesion in Maffucci syndrome and ollier's disease are nearly similar, but it is important to differentiate between the two because there is a greater risk of developing non-skeletal malignancies in Maffucci syndrome (1). Chondrosarcoma is the most frequent musculoskeletal malignancy encountered in Maffucci syndrome, although malignant transformation of the skeletal lesions to fibro sarcoma and vascular lesion to hemangioendothelioma or lymphangosarcoma has been reported as well (2,3,6,9,**10,11**).

The purpose of this case report is to alarm the radiologist, the orthopedic surgeon, and the dermatologist that the X-ray finding of this rare disease is pathognomonic and that a close follow up and early treatment if necessary is the target of the prevention of malignant transformation that is common in this disease. In poorly equipped diagnostic set-ups, phleboliths seen on simple plain films should be considered as one of the clues to differentiate Maffucci syndrome from ollier's disease.

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