Case Report

Maffucci syndrome with unilateral limb: a case report and review of the literature

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Abstract: Maffucci syndrome is a congenital, non-hereditary mesodermal dysplasia manifested by multiple enchondromas and hemangiomas. It is associated with diverse secondary musculoskeletal deformities, which is exceedingly rare. We report a case of hemangiomas and enchondromas localized in the unilateral limb in a patient with Maffucci syndrome. Treatment consists of orthopedic and surgical intervention to minimize deformities and for cosmetic purpose. Careful surveillance for malignant degeneration of both skeletal and non-skeletal tumors, especially in the brain and abdomen, is essential.

Key Words: Maffucci syndrome; enchondromatosis; hemangiomas; unilateteral limb

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Introduction

Maffucci syndrome is an exceedingly rare clinical entity characterized by multiple enchondromas and hemangiomas. It consists of combined occurrence of multiple enchondromas and vascular tumors (1-4). Enchondromas are benign cartilaginous tumors that may develop in any site, but are most frequently found in phalanges and long bones. Complications include spontaneous fracture through area of advanced rarefaction in 26% and sarcomatous degeneration of enchondroma in 15-30%. Chondrosarcomatous transformation occurs in approximately 30% to 40% of enchondromas (2). Hemangiomas are most frequently located in the dermis and subcutaneous fat adjacent to areas of enchondromatosis or elsewhere, and may show a similar distribution with regard to laterality.

Case report

A 33-year-old female born of a noninbred marriage presented with multiple soft tissue swellings of right hand and foot, thigh and lower limb since the age of 13 years. Physical examination showed bony painless swellings on the phalanges of right foot (*Figure 1*) and multiple hemangiomas of over right side of right hand and foot, thigh and lower limb. Examinations of her parents were normal.

Radiographs showed multiple well-defined, irregularly expanded and radiolucent lesions corresponding to intra-osseous benign cartilaginous tumors or enchondromas, localized in the second phalange of the third and fourth right digits. Whole body bone scan showed multiple hot spots in right humerus, femur, tibia and knee. Then skeletal survey showed multiple enchondromas on right humerus, femur, pelvis, tibia and fibular head (*Figures 2,3*).

The patient underwent complete removal of visible hemangioma at right foot. In the operation, the phalanx cortexes of the 3rd and 4th toes were found erosion with some wax-like substance (*Figure 4*). And we conducted bone biopsy of right fibular head. The tumors of right toes and fibular head were waxy, white, avascular, which could not be completely dissected away from the cortex.

The patient was finally discharged uneventfully. The pathological study proved a mature chondroma. The tumors of right toes and fibular head were accompanied by benign

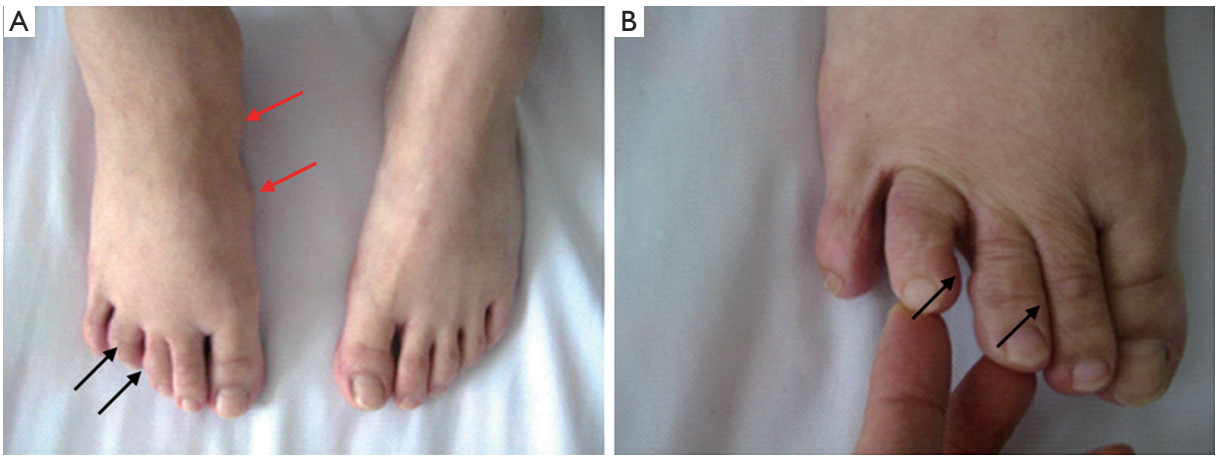

Figure 1 Right toes were deformed due to enchondromas (black arrow), seen hemangiomas (red arrow) below the medial malleolus (A). Clinical symptoms appeared in the right side of the 3rd and 4th toes (B)

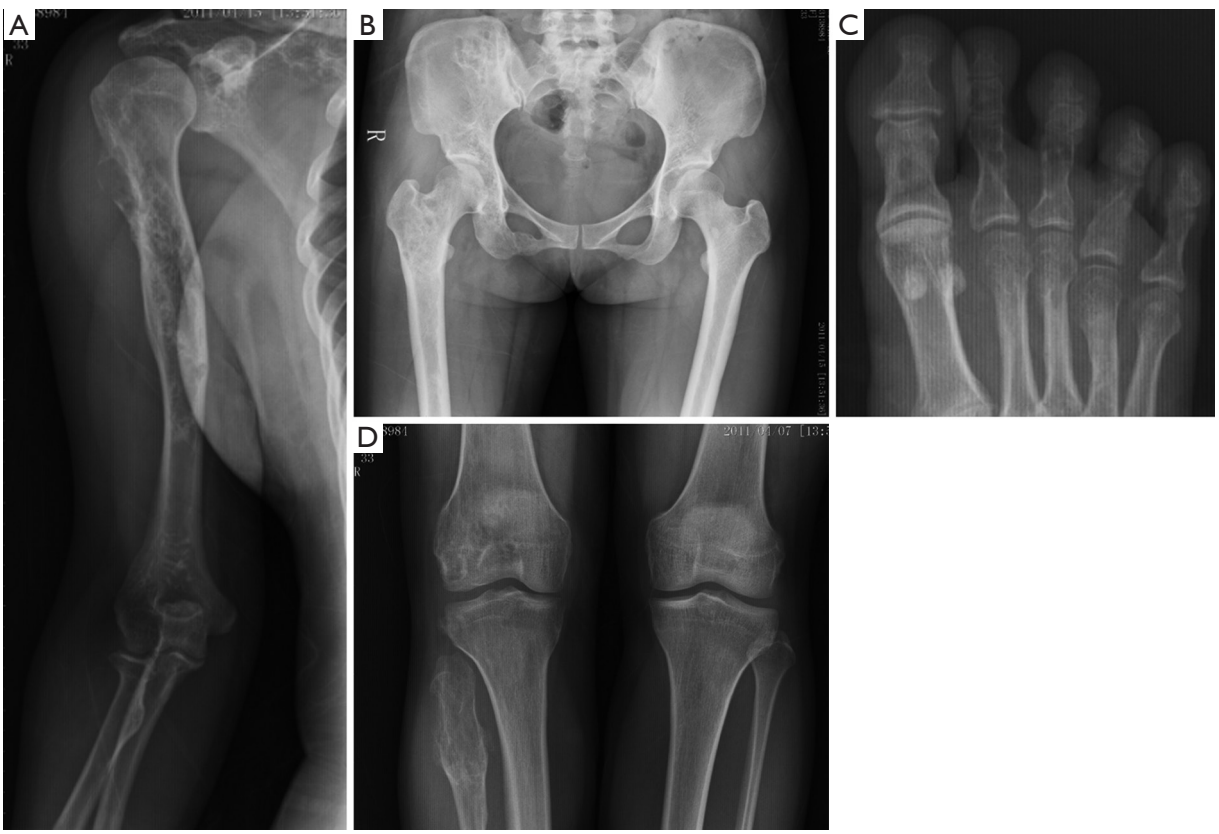

Figure 2 The right scapula bone density of local uneven, the right humerus (A), the right iliac wing and ischial (B), showing multiple cystic lucent zones with irregular bone sclerosis around them; C. Proximal phalanx structure of right foot was unclear, some of which showed thinning slightly bulging, less uniform density, and some small cystic changes; D. Right distal femur and right fibula bone segment with irregular and uneven density, showing multiple cystic in low-density, rough and irregular cortical bone, local swelling change. There was some irregular sclerosis around them

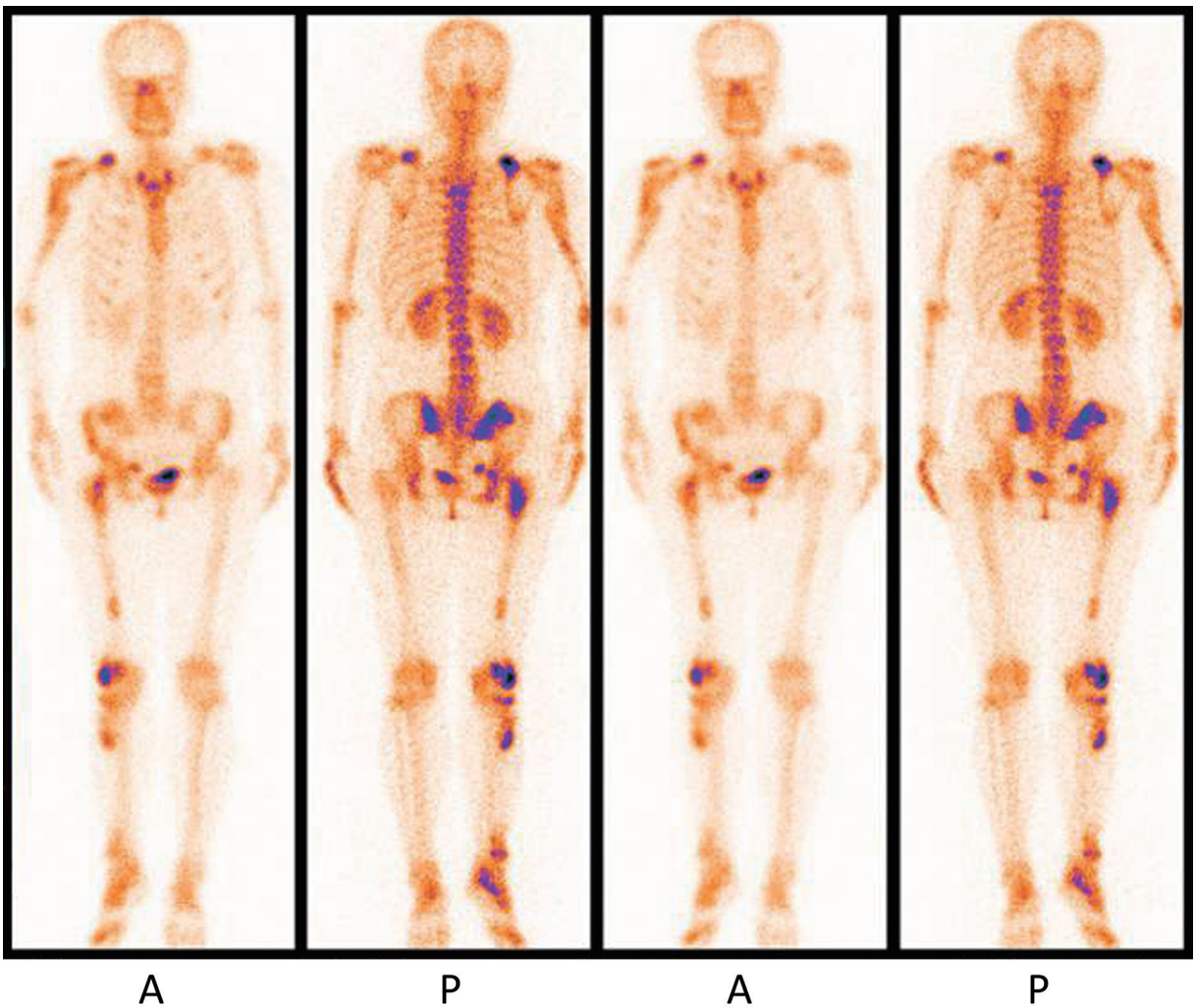

Figure 3 Whole body bone scan demonstrated abnormal uptake signal in the right humerus, scapula, pelvis, right femur, and right fibula in various locations. A, anterior; P, posterior

hemangioma of right foot (*Figure 5*). Follow-up of the patient has revealed no signs of malignant transformation or recurrence (*Figure 6*).

Discussion

Maffucci first reported the syndrome in 1881, and named it as Maffucci syndrome. Multiple enchondromas are complicated by multiple hemangiomas. The disease occurs in unilateral side of the body, and more commonly in the hand, foot and forearm. The reason is unknown, and there is no difference in incidence among races, families, genders or genetics (5,6) in children or youth.

In this syndrome, all the cartilage of the bone can be involved, commonly in the metacarpal and phalanx (87.8%). It can be diagnosed relatively easily solely on clinical grounds. Ancillary tests sometimes need to be taken, including radiographs and arteriography (occasionally), especially in large tumors to determine arterial embolization.

Enchondromas exists not only most frequently at the small bones of the hands and feet, the long tubular bones, but also the flat bones, such as pelvis. Enchondromas are usually in close proximity to or in continuity with growth plate cartilage. Consequently, they might be the result from abnormal regulation of proliferation and terminal differentiation of chondrocytes in the adjoining growth plate. The osseous lesions most frequently involve the phalanges, metacarpals and metatarsal. Maffucci syndrome might be associated with three types of vascular lesions: cavernous hemangiomas, phlebectasias and

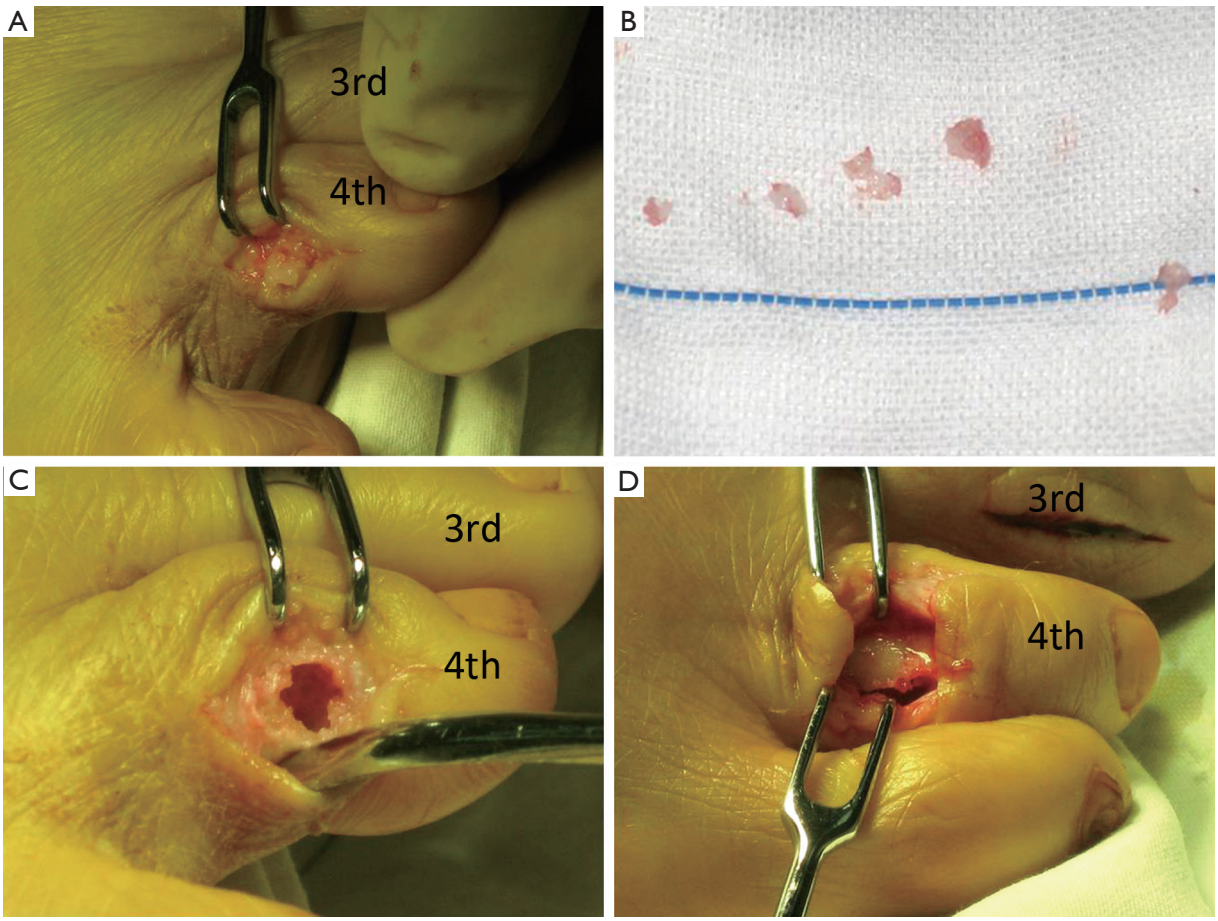

Figure 4 In the operation, the phalanx cortexes of the 3rd and 4th toes were found erosion (A) with some wax-like substance (B); C. Scrape of the lesions; D. implant allogeneic bone; 3rd, the third toe; 4th, the forth toe

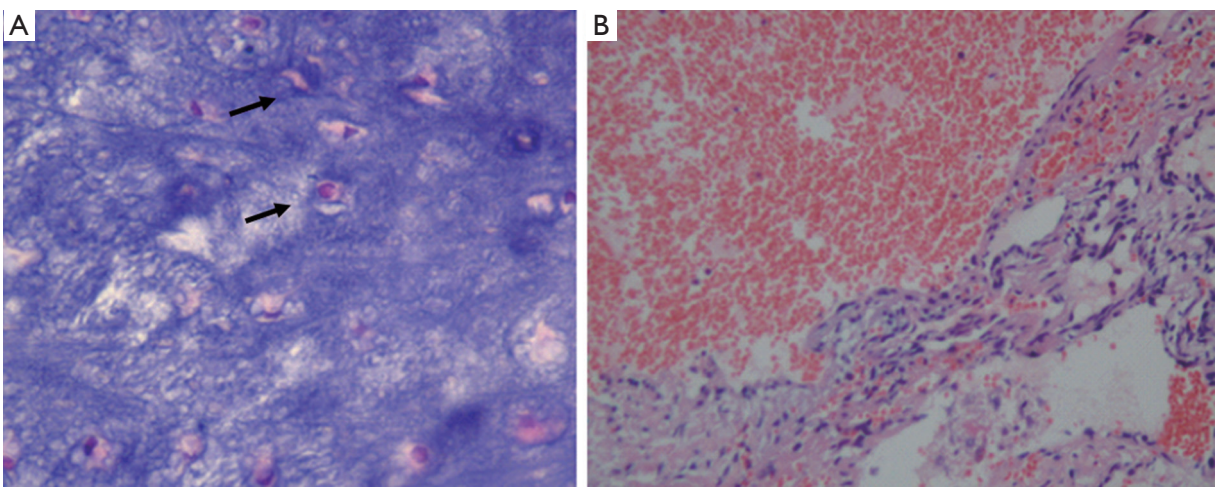

Figure 5 Microscopical features of the pathological sections of tumor. A. Hyaline cartilage (arrows), irregular lobulated calcification within the cartilage matrix and calcified debris (Alcian Blue staining, ×80); B. Focal endothelial cells, vascular epithelial cells, fibroblasts and mast cells and other components, in line with the performance of hemangioma (Haematoxylin-Eosin staining, ×80)

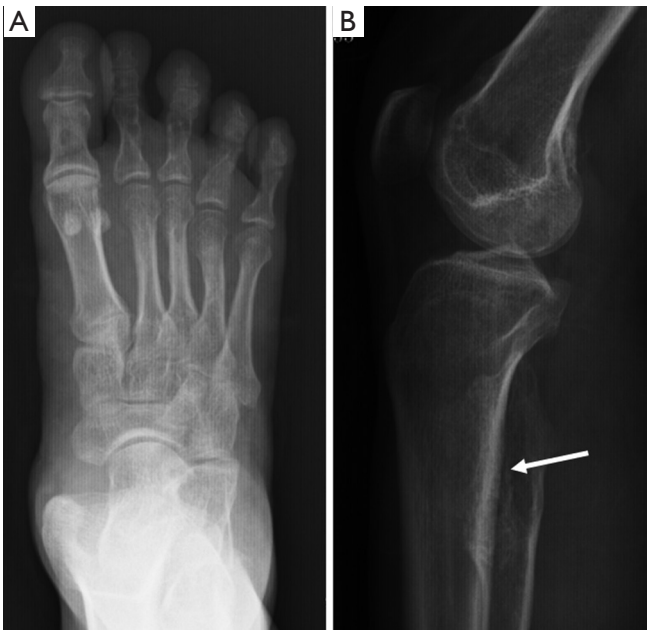

Figure 6 The X-ray film of post-operation showed lesion curettage and bone implants (A), and the scrapings of a part of the fibular head (B, white arrow) at the 3rd and 4th toes

lymphangiectasias-lymphangiomas (6). Clinical problems caused by enchondromas include skeletal deformity and the potential for malignant change, reported in approximately 30% of reported cases.

All enchondromas and hemangiomas of this patient were located in right limbs. All of them were asymptomatic and had no obvious relationship with each other. X-ray showed they are irregularly shaped. Radiolucent areas were found with no stippled calcification within the right humerus, femur, tibia and fibular head right scapular and pelvic dyschondroplasias, while there was no phlebolith in soft tissue.

If absence of clinical symptoms or findings, no treatment is needed (7,8). Surgery is indicated only in the case with complications, such as pathological fractures, growth defects and malignant transformation (7,8). The goal of surgery is to remove the tumor mass and make histological diagnosis. If necessary, chondrosarcoma needs adjunctive therapies (7), including sclerotherapy, irradiation and laser therapy. In this case, the patient felt uncomfortable because of the swellings of the enchondromas and hemangiomas on right foot. Therefore, it was appropriate to only excise these masses. Moreover, tissue biopsy of right fibular head was taken, which showed benign enchondromas and hemangiomas.

In conclusion, when the orthopedic surgeon discovered the patients with multiple enchondromas by X-ray, they should be examined carefully to determine whether hemangiomas exist. Treatment for Maffucci syndrome should be aimed at early detection of malignant transformation as well as symptom relief. The key point of follow-up is to master the progress of multiple hemangiomas in the internal organs, then take biopsy and make pathologic conclusion.

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