Bone Scintigraphy Findings of A Case with Maffucci's Syndrome

Sang Mi Lee · Jong Jin Lee · Yu Kyeong Kim ·
Jeong Won Lee · Jin-Haeng Chung · Sang Eun Kim

Received: 30 November 2009 / Revised: 2 February 2010 / Accepted: 16 February 2010 / Published online: 21 April 2010
© Korean Society of Nuclear Medicine 2010

Abstract Maffucci's syndrome is a sporadic rare congenital disease that is characterized by enchondromatosis and soft tissue hemangiomas. A systemic evaluation should be considered because this syndrome is related to generalized mesodermal dysplasia, which has a high likelihood of a malignant transformation. Whole-body bone scintigraphy might be helpful for detecting skeletal involvement. We present a case of Maffucci's syndrome using bone scintigraphy to evaluate the extent of the disease.

Keywords Maffucci's syndrome · Bone scintigraphy · Enchondromatosis

Introduction

Maffucci's syndrome is a rare congenital nonhereditary disease with mesodermal dysplasia manifesting as enchondromatosis, secondary malignancy, and mesenchymal dysplasias. Mesenchymal dysplasias include subcutaneous hemangioma, phlebolitis, and less often, lymphangiomas [1]. The risk of a malignant transformation is high in patients with Maffucci's syndrome. A

sarcomatous transformation might be seen in bone and soft tissue lesions but it is found more frequently in enchondromas. Because this syndrome is characterized by generalized mesodermal dysplasia, whole-body bone scintigraphy can be helpful in identifying the extent of bone involvement. This paper describes the bone scintigraphy findings in a patient with Maffucci's syndrome who presented with a chondrosarcoma.

Case Report

A 45-year-old female patient visited the orthopedist at our medical center complaining of right-knee-joint pain for a 3-week duration. Her family history and previous medical history were unremarkable. Upon a physical examination, swelling and effusion was observed at the right knee joint area. On the physical exam, multiple, rubbery, and variously sized (less than 3 cm) nodules were palpable on the feet. Her neurological examination was normal. The knee series of plain radiographs revealed an osteolytic lesion in the left proximal fibula (Fig. 1). However, no significant finding was shown in the right-knee joint. Also, right-knee pain subsided after medication with a non-steroid anti-inflammatory drug (NSAID).

A magnetic resonance imaging (MRI) scan was performed to evaluate the left fibula lesion. On the T2-weighted MRI images, multiple variable-sized expansile circumscribed lobulated high-signal intensity mass lesions were observed at the left distal femur, proximal tibia and fibula. Short T1 inverse recovery (STIR) images showed high signal intensity in the marrow around the mass lesions (Fig. 2). These MRI findings were suggestive of enchondromas with malignant transformation. Other plan radiography demonstrated multiple variable-sized expansile

S. M. Lee · J. J. Lee (✉) · Y. K. Kim · J. W. Lee · S. E. Kim
Department of Nuclear Medicine,
Seoul National University College of Medicine,
Seoul National University Bundang Hospital,
300 Gumi-dong, Bundang-gu,
Seongnam-si, Gyeonggi-do 463-707, Korea
e-mail: jjwalking@naver.com

J.-H. Chung
Department of Pathology,
Seoul National University College Medicine,
Seoul National University Bundang Hospital,
Seongnam-si, Korea

Fig. 1 Radiograph (anteroposterior view) of both knee joints revealed a radiolucent lesion of the left fibula (*arrow*)

osteolytic lesions in the phalanges of the feet, both metatarsal bones, distal tibias, and right fifth phalanges of the hands, suggesting enchondromatosis (Fig. 3). In addition, numerous phleboliths were observed at the feet.

^{99m}Tc -hydroxymethylene diphosphonate (HDP) bone scintigraphy performed to determine the extent of the disease revealed focal increased uptake in both tibias, femurs, left scapula and left pelvic bone (Fig. 4). Of these lesions, the left scapular, both ankle joints and left pelvic

bones lesions were clinically unknown and were newly found. MRI scans focusing on these lesions were performed for further workup of the new lesions at the left scapular, both ankle joints and left pelvic bone lesions. On the T2-weighted MRI images, multiple variable-sized lobulated masses with high signal intensity were observed at the left shoulder, ankle and hip joint area. These findings corresponded to the lesions on bone scintigraphy.

There was no evidence of other malignancies in other studies, such as chest computed tomography (CT) and abdominal ultrasonography. Incisional biopsies of the left proximal fibula, left tibia shaft and subcutaneous nodule of feet were performed. The resected specimen of the left fibula and tibia revealed a low-grade chondroid neoplasm with permeation of the bone marrow, necrosis and slightly increased cellularity, suggesting chondrosarcoma, grade 1. Furthermore, the specimen of the subcutaneous nodules of feet was a capillary hemangioma consisting of anastomosing vascular channels of capillary caliber (Fig. 5).

Discussion

Maffucci's syndrome was first described in 1881 and approximately 200 cases have been reported thus far. Maffucci's syndrome is characterized by enchondromatosis with a dermal or subcutaneous hemangioma. The differential diagnosis is Ollier's disease. Ollier's disease also shows enchondromatosis but not hemangioma [2]. It was also reported that Maffucci's syndrome has a higher risk of malignant transformation than Ollier's disease [3, 4].

Fig. 2 Coronal T2-weighted MRI images of the left knee joint shows hyperintense masses in the left tibia (**a**, *arrow*) and left fibula (**b**, *arrow*). STIR image shows high signal intensity in the marrow (*arrow*) around the masses in the left tibia and fibula (**c**)

Fig. 3 Plain radiograph shows multiple radiolucent lesions in both phalanges of feet (**a**, *arrow*) and right fifth phalange of the hand (**b**, *arrow*). Numerous phleboliths were observed at both feet

Fig. 4 ^{99m}Tc -HDP bone scintigraphy showed multiple increased uptakes in both tibias, femurs, left scapula, both ankles and left pelvic bone. Bone scintigraphy detected clinically unknown lesions of the left scapula (*arrow*), both ankles (*arrow*) and left pelvic bone (*arrow*)

The affected bone lesions in Maffucci's syndrome are in the order of prevalence, the hand, foot, tibia, fibula, femur, radius, ulna, humerus, ribs, scapula, pelvis, vertebra, skull, clavicle and sternum [1]. The most common malignant transformation of enchondromatosis is chondrosarcoma [1]. On plain radiography, findings such as endosteal or cortical erosion, frank cortical destruction and an associated soft tissue mass extending beyond the cortical disruption in the long bones are indicative of a malignant transformation [5]. The typical MRI findings of enchondroma and chondrosarcoma are high signal intensity on the T2-weighted images [6]. It is sometimes insufficient to differentiate enchondroma from chondrosarcoma using the T2-weighted images. Abnormal peritumoral and soft tissue signal on the STIR images can help differentiate chondrosarcoma from enchondroma [7]. The MRI findings in our case also showed the same findings as previous reports.

Unfortunately, bone scintigraphy has a difficulty in differentiating chondrosarcoma from enchondroma. Even repeated bone scan failed to reveal a malignant transformation of enchondromas [8, 9]. Therefore, bone scintigraphy appears to have a limited role in detecting a malignant transformation of enchondromatosis. Instead, bone scintigraphy might be useful for determining the extent of the disease and detecting new bone lesions. Karacalioglu et al. [10] reported that bone scintigraphy and RBC imaging might be helpful in diagnosing the extent of the disease. Other studies also revealed the excellent role of bone scintigraphy for determining disease extent [11, 12]. In this study, unexpected left scapula, ankle joint and hip joint lesions were detected by bone scintigraphy, which suggests

Fig. 5 (a) The resected specimen of the left fibula and tibia showed well-differentiated chondrosarcoma (grade 1) with mild cytologic atypia (hematoxylin and eosin, $\times 200$). (b) The specimen of the

subcutaneous nodules of feet showed capillary hemangioma consisting of network of neoplastic capillaries (hematoxylin and eosin, $\times 400$)

that bone scintigraphy might be useful for evaluating the extent of the disease.

References

- Lewis RJ, Ketcham AS (1973) Maffucci's syndrome: functional and neoplastic significance. Case report and review of the literature. *J Bone Joint Surg Am* 55:1465–1479
- Silve C, Juppner H (2006) Ollier disease. *Orphanet J Rare Dis* 1:37
- Schwartz HS, Zimmerman NB, Simon MA, Wroble RR, Millar EA, Bonfiglio M (1987) The malignant potential of enchondromatosis. *J Bone Joint Surg Am* 69:269–274
- Mellon CD, Carter JE, Owen DB (1988) Ollier's disease and Maffucci's syndrome: distinct entities or a continuum. Case report: enchondromatosis complicated by an intracranial glioma. *J Neurol* 235:376–378
- Sun TC, Swee RG, Shives TC, Unni KK (1985) Chondrosarcoma in Maffucci's syndrome. *J Bone Joint Surg Am* 67:1214–1219
- Unger EC, Kessler HB, Kowalyszyn MJ, Lackman RD, Morea GT (1988) MR imaging of Maffucci syndrome. *AJR Am J Roentgenol* 150:351–353
- Janzen L, Logan PM, O'Connell JX, Connell DG, Munk PL (1997) Intramedullary chondroid tumors of bone: correlation of abnormal peritumoral marrow and soft-tissue MRI signal with tumor type. *Skeletal Radiol* 26:100–106
- Adams BK (1989) Scintigraphy in benign bone tumours. A report of 4 cases. *S Afr Med J* 76:112–114
- Fallahi B, Bostani M, Gilani KA, Beiki D, Gholamrezanezhad A (2009) Manifestations of Ollier's disease in a 21-year-old man: a case report. *J Med Case Reports* 3:7759
- Karacalioglu O, Gunalp B, Ilgan S, Arslan N, Ozguven MA (2004) Combined Tc-99 m MDP and Tc-99 m labeled RBC imaging in a case of Maffucci's syndrome. *Clin Nucl Med* 29:817–820
- Trikha V, Gupta V, Kumar R (2003) Ollier's disease: characteristic Tc-99 m MDP scan features. *Clin Nucl Med* 28:56–57
- Gruning T, Franke WG (1999) Bone scan appearances in a case of Ollier's disease. *Clin Nucl Med* 24:886–887