Clinical Case Report

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Multiple unexpected lesions of metachondromatosis detected by technetium-99m methylene diphosphonate SPECT/CT

A case report

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Abstract

Rationale: Metachondromatosis (MC) is a very rare genetic disease, which is infrequently reported worldwide, which leads to osteochondroma and enchondromatosis. The disease has been shown to be associated with loss of function of the tumor suppressor gene "protein tyrosine phosphatase, non-receptor type 11" (*PTPN11*).

Patient concerns: A 12-year-old female was admitted to the hospital with pain due to an enlarged mass in her left fifth finger.

Diagnosis: Examination of the left hand by computed tomography (CT) revealed an expanding type of round and low-density lesion in the fifth proximal phalanx. The patient then underwent technetium-99m methylene diphosphonate single-photon emission CT/CT (^{99m}Tc-MDP SPECT/CT) to assess the nature of the lesion. The SPECT/CT image revealed dilated osteopathy and increased activity of the fifth proximal phalanx on the left hand. Unexpectedly, the examination of the right hand revealed slight expanded lesions and increased activities of the third metacarpal and proximal phalange, as well as the fourth proximal phalange and the middle phalanx. On the basis of the patient's symptoms and the results of the above-mentioned examinations, we diagnosed the patient as having MC in her hands.

Intervention: Considering the pain of the fifth finger of the left hand, the patient underwent debridement of the fifth proximal phalanx of the left hand and internal fixation with bone graft taken from the body.

Outcomes: The patient was discharged after a week of observation. One year later, she was admitted to the hospital again for removal of the bone healing internal fixation after osteoma surgery. Preoperative ^{99m}Tc-MDP SPECT/CT revealed that the left-handed lesions displayed postoperative changes, while the multiple lesions in the right hand increased in volume but remained unchanged in number.

Lessons: This case revealed the CT and ^{99m}Tc-MDP SPECT/CT imaging features of MC. Specifically, SPECT/CT imaging contributed to the diagnosis of clinically asymptomatic bone lesions, and the 3D SPECT/CT fusion allowed a more comprehensive and intuitive view of the lesion by combining anatomy and function.

Abbreviations: 3D = 3-dimensional, ^{99m}Tc-MDP SPECT/CT = ^{99m}Tc-methylene diphosphonates single-photon emission computed tomography/computed tomography, CT = computed tomography, MC = metachondromatosis.

Keywords: ^{99m}Tc-MDP SPECT/CT, CT image, metachondromatosis

1. Introduction

Metachondromatosis (MC) is a rare disorder characterized by the presence of both enchondromas and osteochondromas in children.[1,2] It is an autosomal dominant disease, which has been shown to be associated with *PTPN11* gene dysfunction.[1,3] MC-related osteochondromas are mainly located in the hands and feet,[4] primarily distributed to the toes and pointing to adjacent growth plates.[3,4] Osteochondromas result from abnormal cartilage growth, also known as hamartoma. A typical tumor consists of a cap of cartilage and a bone tissue protruding from the side of the bone.[1,5,6] Compared with the enchondromas in patients with enchondromatosis, the enchondromas of MC patients mainly affect the iliac and metaphysics of long bones,[7] and vascular necrosis is one of their common complications.[8,9] The incidence of MC is thought to be less than 1 in 1,000,000[10]; fewer than 100 cases have been reported in the literature.

2. Case report

A 12-year-old female with a lump on the fifth finger of the left hand for 2 years and progressive pain for 1 year was admitted to our hospital on January 17, 2017. Before admission, she underwent an X-ray examination of her left hand at a community hospital, showing a rounded osteophyte in the fifth proximal phalanx. Unfortunately, her X-ray image was not available to us. The initial computed tomography (CT) image of the left hand

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The authors report no conflicts of interest.

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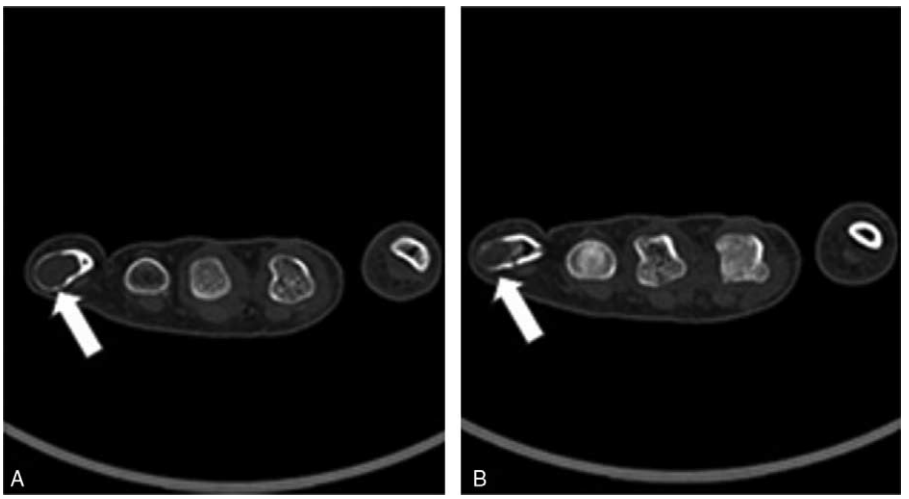

Figure 1. The initial CT image of the left hand obtained showed the presence of a low-density tumor of approximately 1.0 × 1.3 cm in the fifth proximal phalanx, on the left side where the cortical bone was dilated and thinned.

obtained at our hospital showed the presence of a low-density tumor of approximately 1.0 × 1.3 cm in the fifth proximal phalanx, on the left side where the cortical bone was dilated and thinned (Fig. 1 arrows). The clinician advised the patient to undergo a SPECT/CT to determine whether there were similar lesions in her other bones. The day after admission, she underwent a SPECT/CT examination. The technetium-99m methylene diphosphonate single-photon emission CT/CT (^{99m}Tc-MDP SPECT/CT) image revealed dilated osteopathy and increased activity of the fifth proximal phalanx on the left side (Fig. 2: the solid bold arrows on B, C, D, and E). In addition, unexpectedly, examination of the right hand revealed slight expanded lesions and high MDP uptake in the third metacarpal (Fig. 2: the hollow thick arrows on B, C, F, and G) and proximal phalanges (Fig. 2: the long arrows on B, C, H, and I), as well as the fourth proximal phalanges (Fig. 2: the short arrows on B, C, J, and K) and the middle phalanx (Fig. 2: the thin arrows on B and C). It was obvious that the above lesions can be observed more intuitively by 3D SPECT/CT fusion image (Fig. 2C). Fortunately, no high MDP uptake was found in other bones, such as that femur, tibia, and pelvis. Her left hand lesion looked like an osteochondroma, and her right hand lesions looked more like multiple enchondromas. On the basis of the above findings, we have reason to suspect that this girl had MC.

Considering the pain in the fifth finger of the left hand, the patient underwent debridement of the fifth proximal phalanx of the left hand and internal fixation with bone graft taken from the body on the fourth day. Subsequent pathological analysis revealed that the surgical specimen contained a hyaline cartilage cap (Fig. 3: long arrow) and underlying mature lamellar bone with marrow elements (Fig. 3: short arrow), which was microscopically characterized by the presence of an osteochondroma (no fibrous perichondrium outer layer was found in this specimen due to sectioning). In addition, an area of endochondral ossification can be observed between these 2 areas (Fig. 3: asterisk). As no clinical symptoms, such as pain, manifested due to the lesions in the right hand, no treatment was required for these lesions.

On January 28, 2018, the patient was readmitted to the hospital for removal of the bony union internal fixation after the surgical removal of the osteochondroma of the fifth finger of the left hand. She first underwent pre-surgery ^{99m}Tc-MDP SPECT/CT. The SPECT/CT image revealed that the titanium plate on the fifth proximal phalanx of her left hand (Fig. 4: the solid bold arrows on B, C, D, E, and F) did not fall off or break; the number of multiple lesions (the lesion in the third metacarpal: the hollow thick arrows on G, H, I, and J; the lesion in the third proximal phalanges: the long arrows on K, L, M, and N; the lesion in the fourth proximal phalanges: the short arrows on M, N, O, and P) in her right hand did not change compared with the previous examination a year ago, but clearly expanded while the cortical bone became thinner at the corresponding position and showed high MDP uptake. No abnormal uptake of MDP was found in other bones of the body. The patient reported slight pain in the left hand metacarpal bone position but refused treatment.

3. Discussion

MC is a rare form of exostosis and enchondromatosis tumor syndrome. It was first reported by Maroteaux in 1971,[11] and since then, case reports have been scarce. MC is a genetic autosomal dominant disorder, which recently has been reported to be associated with mutations in the *PTPN11* gene.[1,3] However, our patient did not undergo a genetic analysis for *PTPN11* gene mutations and her parents did not have a similar condition. Thus, this case seems to be different from the vast majority of cases currently reported, which usually have a family history.[1,3]

The syndrome of MC manifests early in childhood. The clinical process of MC is unpredictable, as some lesions may grow simultaneously while others may degenerate. One of the clinical features of MC is the tendency of the osteochondromas to degenerate or disappear after the first or second decade.[10,12] Due to the reduced number of MC case reported in the literature, it is difficult to make establish a formal treatment and monitoring recommendations. The indication of surgical resection is mainly determined by symptomatic manifestation and related to pain and nerve compression.[13] We detected multiple lesions in the patient's hands by ^{99m}Tc-MDP SPECT/CT. The exostosis in her left hand was surgically removed and pathologically determined to be osteochondroma. The multiple lesions in her right hand were not surgically removed, so no pathological analysis was performed to confirm whether the lesions were enchondromas.

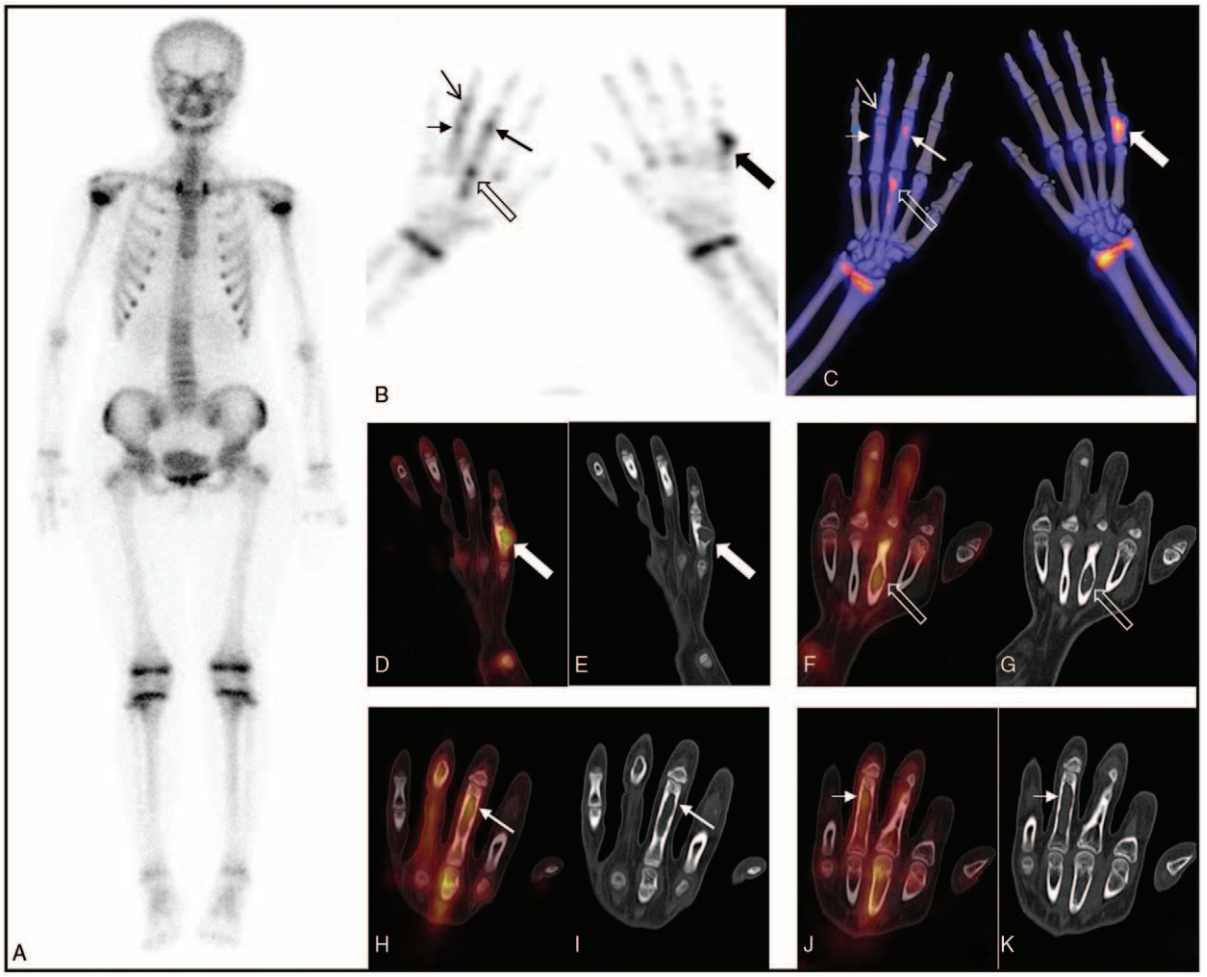

Figure 2. The ^{99m}Tc-MDP SPECT/CT image revealed dilated osteopathy and increased activity of the fifth proximal phalanx on the left side (the solid bold arrows on B, C, D, and E). In addition, unexpectedly, examination of the right hand revealed slight expanded lesions and high MDP uptake in the third metacarpal (the hollow thick arrows on B, C, F, and G) and proximal phalanges (the long arrows on B, C, H, and I), as well as the fourth proximal phalanges (the short arrows on B, C, J, and K) and the middle phalanx (the thin arrows on B and C). It was obvious that the above lesions can be observed more intuitively by 3D SPECT/CT fusion image (C).

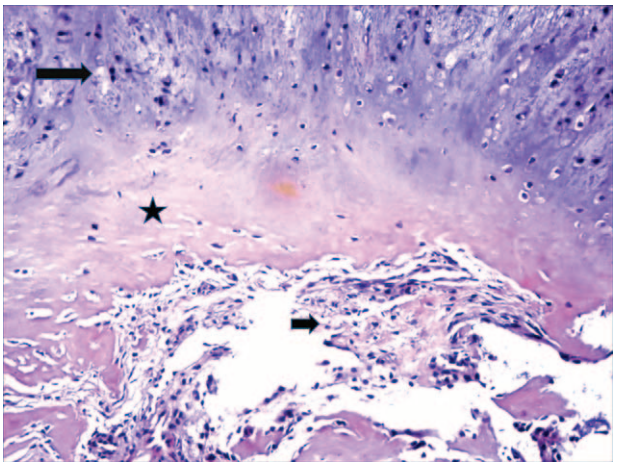

Figure 3. The pathological analysis revealed that the surgical specimen contained a hyaline cartilage cap (long arrow) and underlying mature lamellar bone with marrow elements (short arrow), which was microscopically characterized by the presence of an osteochondroma. In addition, an area of endochondral ossification can be observed between these 2 areas (asterisk).

Accordingly, the diagnosis of this case as MC remains controversial.

Compared with magnetic resonance imaging (MRI), SPECT/CT has the advantages of early and whole-body imaging, which involves the combination of planar imaging and local tomography. Currently, the application of 3D SPECT/CT fusion imaging technology in the diagnosis of bone diseases is gradually increasing. In addition, it can make the application more comprehensive and achieve an intuitive display by fusing anatomy and function. In our case, multiple endogenic lesions were inadvertently detected in the right hand of the patient by ^{99m}Tc-MDP SPECT/CT examination, which led to the consideration that she had a rare disease, such as MC. SPECT/CT, as a whole-body imaging modality, can effectively detect lesions of the whole body without additional imaging examination. Therefore, the application of SPECT/CT as the preferred imaging modality for further examination of X-ray posterior suspected multiple osteodystrophy may serve as a reference for clinicians.

4. Conclusion

This case reports the unintentional detection of a rare disease with features of MC by ^{99m}Tc-MDP SPECT/CT, which allows the

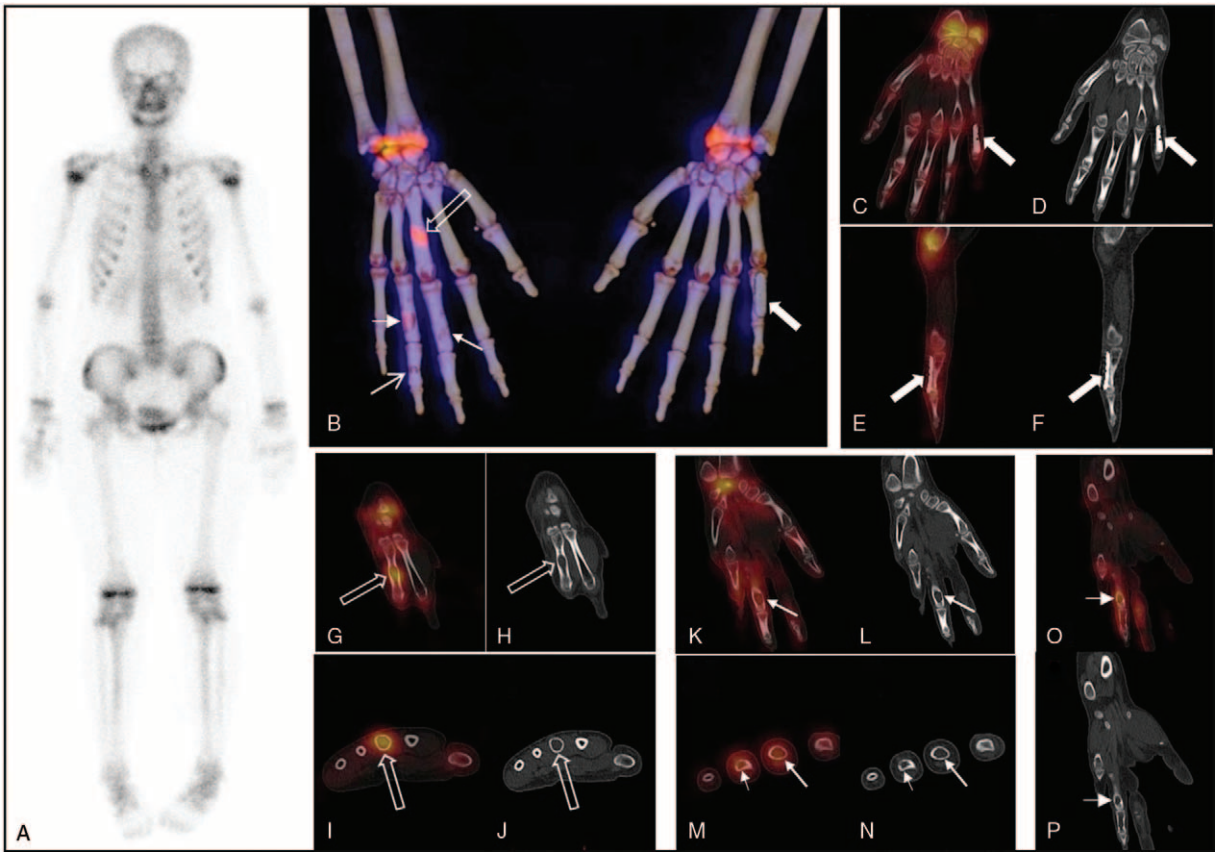

Figure 4. The pre-surgery ^{99m}Tc-MDP SPECT/CT image revealed that the titanium plate on the fifth proximal phalanx of her left hand (the solid bold arrows on B, C, D, E, and F) did not fall off or break. The examination of the right hand revealed expanded lesions and high MDP uptake in the third metacarpal (the hollow thick arrows on G, H, I, and J) and proximal phalanges (the long arrows on K, L, M, and N), as well as the fourth proximal phalanges (the short arrows on M, N, O, and P) and the middle phalanx (the thin arrows on B). Although the number of multiple lesions in her right hand did not change compared with the previous examination a year ago, but clearly expanded while the cortical bone became thinner at the corresponding position and showed high MDP uptake.

visualization and comprehensive of occult lesions though 3D fusing imaging by combining anatomy and function. This suggests that SPECT/CT, as an imaging modality for the detection of occult bone disease, has excellent diagnostic value and application prospect.

4.1. Consent

Informed written consent for the publication of our case report and related images was signed by the patient.

Author contributions

Data curation: Yuting Zou, Yu Chen
Writing – original draft: Zi Wang.

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