Osteopoikilosis found incidentally in a 17-year-old adolescent with femoral shaft fracture

A case report

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Abstract

Rationale: Osteopoikilosis is a rare and asymptomatic disease of the bone, which is often discovered occasionally on radiography for irrelevant complaints. Characterized by multiple, small, circular, or oval-shaped radiodense lesions, it may be confused with bony metastatic tumors.

Patient concerns: The present study describes a case of a 17-year-old adolescent who suffered from pain and movement limitation of his left thigh following a fall from standing height.

Diagnoses: Plain radiographs showed spiral fracture in left femoral shaft; besides, multiple scattered sclerotic lesions of variable size were also observed over the bilateral proximal femurs, left distal femur, proximal tibia, and distal tibia and fibula through X-rays, computed tomography, and magnetic resonance imaging. The patient was finally diagnosed with left femoral shaft fracture and osteopoikilosis.

Interventions: The patient underwent reduction and internal fixation with intramedullary nail a week after injury.

Outcomes: The patient was discharged without any complications 12 days after the surgery. At the 3-month follow-up, the patient recovered well and remained symptom-free with no changes to his sclerotic lesions.

Lessons: Although this case is not so complicated, we have to be cautious when differentiating osteopoikilosis and bony metastases in clinical practice in future, which should avoid causing undue distress to both the patients and doctors.

Abbreviations: CT = computed tomography, MRI = magnetic resonance imaging.

Keywords: autosomal dominant, intramedullary nail, osteopoikilosis, sclerotic lesions, surgery

1. Introduction

Osteopoikilosis or “spotted bone disease” is a peculiar, benign, autosomal dominant disorder characterized by circular or oval-shaped sclerotic bone lesions most commonly involving the hands, feet, pelvis, and ends of long bones.[1,2] No gender difference was observed at all ages with an estimated prevalence of 1 in 50,000 individuals.[3] Although an autosomal dominant inheritance pattern has been reported in certain cases, the etiology and pathogenesis of osteopoikilosis remain elusive.[4] This disease, although benign, is likely to be mixed up with bony metastases. For example, it has been reported that bony metastases from patients with breast cancer have been missed on account of the presence of osteopoikilosis.[5] Thus, we must be prudent to distinguish between osteopoikilosis and metastatic bone tumor in clinical practice.

In the present study, we describe a rare case osteopoikilosis found accidentally in a 17-year-old adolescent who suffered acute left femoral shaft fracture.

2. Presenting concerns

A 17-year-old Chinese adolescent, who was a student, presented to the emergency department complaining of pain and restricted movement over his left thigh for nearly 6 hours following a fall from standing height. The pain was a kind of sharp pain. The pain aggravated when he moved the left lower extremity.
was single. In the general physical examination, no radiation pain was found in the left lower limb. The skin around the middle of the left thigh was bruised and swollen. No neurologic signs or symptoms were observed in the left extremity. Besides, he could not move his left limb due to the extraordinary pain. No fever or respiratory embarrassment accompanying with the injury was observed. Recently, he did not undergo obvious weight loss or expose to tuberculosis.

4. Diagnostic focus and assessment

Further physical examination showed no palpable head, neck, supraclavicular, axillary, epitrochlear, or inguinal lymph nodes. The erythrocyte sedimentation rate (32 mm/h) was a little higher beyond the normal range (normal ranges: 0–20 mm/h). Plain radiographs showed spiral fracture in left femoral shaft; besides, multiple scattered sclerotic lesions of variable size were also observed over the bilateral proximal femurs, left distal femur, proximal tibia, and distal tibia and fibula through X-rays, computed tomography (CT), and magnetic resonance imaging (MRI) (Fig. 1). Consequently, diagnosis of spiral fracture in left femoral shaft and osteopoikilosis was made by an experienced radiologist considering the medical history, symptoms, and imaging findings.

5. Therapeutic focus and assessment

After excluding any surgical contraindications, experienced surgeons conducted the operation to repair the fracture in the left femoral shaft. The patient was placed in a supine position, and following epidural anesthesia, the left thigh was covered with sterile drapes and positioned to expose the surgical field. On the basis of the fluoroscopic imaging, surgeons made the 2 distal nail holes appeared as a perfect circle when inserting distal screws. Then, an appropriate skin incision was made through the soft tissue to the bone cortex. The drill bit and the distal locking hole stayed on the same axis (Fig. 2). The screw insertion was performed when the hole was contacted with the tip of the screw. A wound drainage tube was inserted, and each layer of tissue was sutured to achieve complete hemostasis. The estimated total loss of blood was 150 mL, and no blood transfusion was required during the surgery.

6. Follow-up and outcomes

The patient was discharged without any complications 12 days after the surgery. At the time of the 3-month follow-up, the patient reported no pain or discomfort in his left thigh with no changes to the sclerotic lesions on radiograph. Table 1 summarized the timeline of the patient.

7. Discussion

Osteopoikilosis is a rare lifelong heritable disease characterized by osteosclerotic dysplasia of bones with intricate etiology and pathogenesis. It is diagnosed by radiographies with characteristic numerous, symmetric, homogeneous, and circular or ovoid bone lesions. Patients are usually asymptomatic with laboratory tests and bone scintigraphy normal in most cases. Osteopoikilosis commonly requires no treatment other than education and reassurance for patients but should be identified to prevent unnecessary invasive testing and distress. In this case, the patient suffered an acute femoral shaft fracture and was diagnosed with osteopoikilosis by accident when he had his left
thigh x-rayed. After reduction and internal fixation with intramedullary nail of left femoral shaft, he had a good recovery during follow-up.

Although generally an asymptomatic incidental finding, osteopoikilosis can be confused with other diseases. The major differential diagnoses include usually metastatic bone tumors, mastocytosis, osteosclerotic myeloma, tuberous sclerosis, and metabolic osteopathies.[7] Nevertheless, the symmetric distribution, the tendency for epiphyseal and metaphyseal involvement, and the uniform size of the lesions are unique for diagnosing osteopoikilosis. Besides, bone scintigraphy of osteopoikilosis is commonly normal, while slightly higher activity of the bone metabolism can be detected sometimes. In contrast, skeletal metastasis and tuberous sclerosis are characterised by asymmetric distribution, usual involvement of axial skeleton, especially spine, common osseous destruction, diversity in size, and positive scintigraphic findings, which means bone scintigraphy is usually crucial for distinguishing osteopoikilosis from primary bone tumors or osteoblastic bone metastases.[8] Our patient did not undergo bone scintigraphy because of typical imaging features of osteopoikilosis.

In conclusion, characterized by circular or oval-shaped sclerotic bone lesions that most frequently involving the hands, feet, pelvis, and ends of long bones, osteopoikilosis is not so difficult for diagnosing. However, it can be mistaken for metastatic bone tumors or other diseases because of similar imaging characteristics, which may result in immoderate invasive examinations and distress to both patient and doctor. Upon most occasions, bone scintigraphy plays a key role in distinguishing osteopoikilosis from metastatic bone tumors.

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Table 1

| Case report timeline. | Details |
|-----------------------|---------|
| Presenting symptoms   | Pain and movement limitation of his left thigh following a fall from standing height |
| First investigations  | Limited motion in the left lower limb; No radiation pain found in the left lower limb; Bruised and swollen skin around the middle of the left thigh; No neurologic signs or symptoms observed in the left extremity |
| Plain radiographs     | Spiral fracture in left femoral shaft; Multiple scattered sclerotic lesions of variable size over the left proximal and distal femur, proximal tibia, distal tibia, and distal fibula |
| CT                    | Multiple sclerotic lesions of different size in the distal femur |
| MRI of left shoulder  | Scattered sclerotic lesions of various size over the bilateral proximal femur |
| Surgical intervention | Reduction and internal fixation of the left femoral shaft with intramedullary nail |
| Postoperative follow-up | Three-month follow-up. Recovered well. Remained symptom-free with no changes to the sclerotic lesions |

CT = computed tomography, MRI = magnetic resonance imaging.