



CASE REPORT

Bilateral Atypical Subtrochanteric Femoral Fractures with Primary and Secondary Bone Healing in Pycnodysostosis

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Abstract

Atypical subtrochanteric femoral fractures are a common problem associated with pycnodysostosis. Pycnodysostosis is a rare sclerotic bone disease caused by a mutation in the cathepsin K gene. Fracture healing in pycnodysostosis cases is typically inferior. Here, we report a case of bilateral atypical subtrochanteric femoral fractures in one patient with pycnodysostosis. The right subtrochanteric fracture was treated with open reduction and internal fixation (open plating), and united through primary bone healing, while the left one was treated with closed reduction and internal fixation (submuscular plating), and united through secondary bone healing. Although the time to bony union was delayed, fracture union after extramedullary osteosynthesis was obtained in both atypical fractures, demonstrating that both primary and secondary bone healing is possible in patients with pycnodysostosis.

Level of Clinical Evidence 4.

Keywords Femur · Fracture · Pycnodysostosis · Bone healing

Introduction

Pycnodysostosis is a rare sclerotic bone disease caused by cathepsin K deficiency [1, 2].

The most two common typical clinical features of pycnodysostosis are short stature, increased bone density. To date, it is difficult to find a full description of clinical manifestations of pycnodysostosis in the literature. Bone resorption is decreased in pycnodysostosis due to cathepsin K deficiency and impaired bone resorption leads to disruption in the bone healing process. Radiological findings of pycnodysostosis

are similar to those of osteopetrosis, a disease that causes increased bone density due to a defect in bone reabsorption. The diagnosis of pycnodysostosis can be confirmed by molecular testing.

Subtrochanteric femoral fractures in patients with pycnodysostosis are similar to atypical subtrochanteric femoral fractures associated with bisphosphonate use [2].

These fractures are often treated with internal fixation using either open reduction or closed reduction, but the impact of this treatment on the healing process is not well understood [3]. Only a few cases of subtrochanteric femoral fractures in patients with pycnodysostosis on the bone union have been reported [1–3]. Here, we report a case of bilateral subtrochanteric femoral fractures in one patient with pycnodysostosis.

This case suggests that fracture healing after extramedullary osteosynthesis in pycnodysostosis can be achieved by either primary or secondary bone healing without bone grafting.

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Case Report

A 40-year-old woman sustained a right subtrochanteric femoral fracture after falling from a bicycle. The patient did not remember the family history well. There was no history of previous fractures. The patient had abdominal organomegaly, which was suspected to account for the ovarian cystadenoma that was found on abdominal computed tomography. Physical examination of the fracture site revealed tenderness over the proximal thigh. There was no open wound. There was no distal neurovascular deficit distally.

The patient was of short stature (150 cm), had abnormal facial features due to micrognathia, which included a large nose and abnormal dentition. Radiographs showed common features of atypical subtrochanteric femoral fracture, including transverse fracture, medial and lateral cortical thickening, and medial cortical spike (Fig. 1). A subsequent skeletal survey revealed an open fontanelle, an obtuse mandibular angle, cortical thickening in all long bones, widening of the distal femur (Fig. 2) (similar to the Erlenmeyer flask deformity), and tapering of the distal phalanges on both feet and hands (Fig. 3).

No hematological abnormalities were observed. The patient had generalized increased bone density, as measured through bone densitometry. *T* scores of the lumbar spines (L1, L2, L3, and L4) were +4.3, +4.3, +5.8, and +6.1, respectively. *Z* scores of the lumbar spines (L1, L2, L3, L4) were +5.1, +5.2, +6.6, and +7.0, respectively.

The patient was diagnosed with pycnodysostosis based on clinical and radiological findings and genetic analysis for cathepsin K mutation. There was an incomplete transverse fracture in the left subtrochanteric area for which the patient refused preventive interventions (Fig. 1).



Fig. 1 Both hip anteroposterior radiograph shows common features of atypical subtrochanteric femoral fractures and a transverse incomplete fracture (arrow) of the left subtrochanter



Fig. 2 Lower extremity radiograph shows long bones' deformities

The right subtrochanteric femoral fracture was secured with fracture site compression using a dynamic hip screw (Synthes, Paoli, PA) without bone graft (Fig. 4a). Patient was followed up monthly for 6 months and then every 3 months thereafter.

Radiographic union developed very slowly, compared to a patient without skeletal abnormalities. Thirty-three months after surgery, the fracture was finally united, as evidenced by the disappearance of the fracture line in at least three cortices (Fig. 4b). The range of motion of both the knee and hip was full.

Six years after the index injury of the right femur, the patient slipped while walking and sustained a left

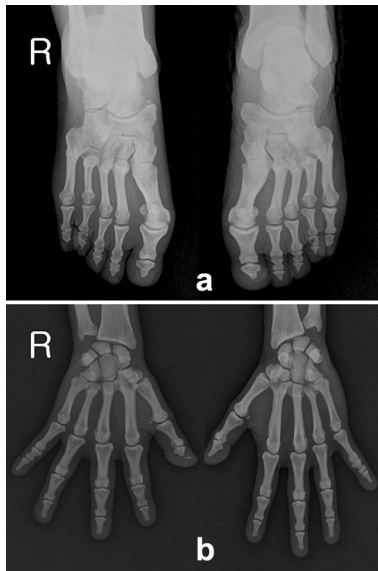


Fig. 3 Both feet (a) and hands (b) radiographs show tapering of the distal phalanges

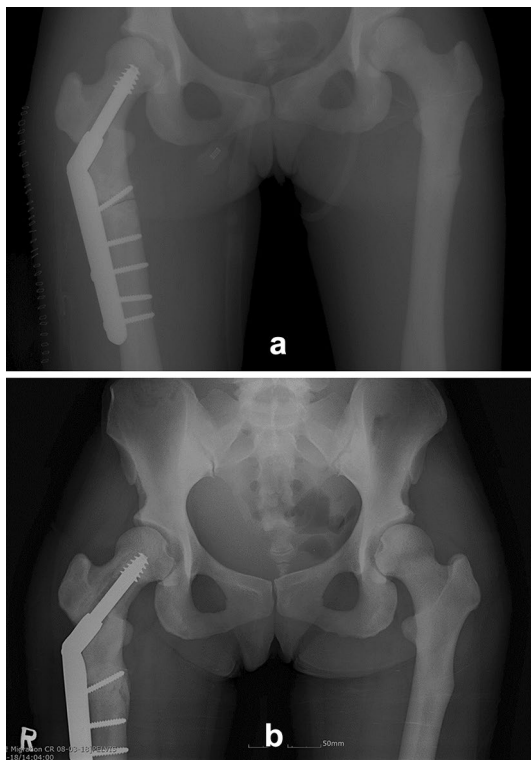


Fig. 4 a The right subtrochanteric femoral fracture was fixed with a dynamic hip screw without bone graft. b Fracture union was finally achieved 33 months after surgery, as evidenced by the disappearance of the fracture line

subtrochanteric femoral fracture. The fracture pattern was very similar to that of the previous right femoral injury (Fig. 5a). The fracture was then fixed with a submuscular plating technique—the minimally invasive plate osteosynthesis (MIPO) technique—with a non-contact-bridging locking plate (Zimmer, Warsaw, IN). A bone graft was not performed.

Again, radiographic union was very slow to develop, similar to the timeframe for the contralateral side. Twenty-four months after surgery, bony union was finally achieved (Fig. 5b). Final follow-up was done at 2 years after surgery (8 years after the first index surgery). The patient could walk without pain. The range of motion of both the knee and hip was full.

Discussion

Subtrochanteric femoral fractures resulting from low-energy trauma are common complications associated with pycnodysostosis [1, 2]. Radiographic findings from these injuries (Fig. 1) are consistent with the atypical subtrochanteric femoral fractures associated with bisphosphonate use [2].

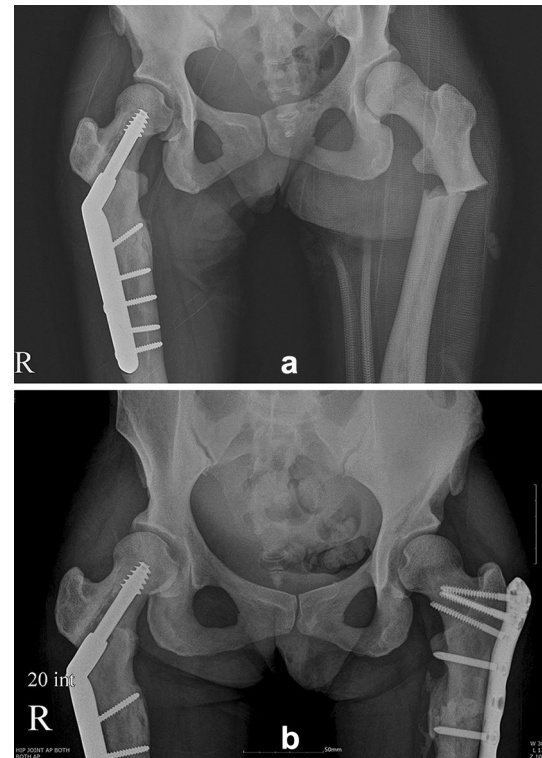


Fig. 5 a The patient also sustained a left subtrochanteric femoral fracture at 6 years after the index injury to her right femur. The fracture patterns for these two injuries were very similar. b The fracture union was finally achieved 24 months after surgery, as evidenced by the formation of bridging calluses

In osteopetrosis, the medullary canal is typically obliterated [4]; however, the medullary canal is open with excessive narrowing by cortical thickening of both the medial and lateral cortices in pycnodysostosis.

Biomechanical stability of intramedullary nailing is superior to that of extramedullary plating. However, some reports described that reaming can be very difficult in pycnodysostosis in the diaphyseal area because the intramedullary canal is narrow due to cortical thickening [3] (Fig. 1).

The fracture healing in pycnodysostosis is delayed because the rate of bone turnover is extremely slow as the osteoclastic function is deficient. In our case, open reduction and internal fixation (extramedullary osteosynthesis for the purpose of primary bone healing) was performed on the right femoral fracture and closed reduction and internal fixation with the MIPO technique (extramedullary osteosynthesis for the purpose of secondary bone healing) was performed on the left femoral fracture.

Ultimately, both fractures progressed to uneventful union, despite time to fracture union being much longer than in normal patients. In this case, it took 33 months (primary, right side) and 24 months (secondary, left side) until radiology confirmed complete bone union. Although we cannot draw any generalizable conclusions from one case report, our case suggests that fracture healing after extramedullary osteosynthesis in pycnodysostosis can be achieved by either primary or secondary bone healing without bone grafting.

Bilateral subtrochanteric femoral fractures associated with pycnodysostosis that achieved bony union by different bone healing mechanisms (primary and secondary bone healing) may be unique in the literature [5]. We believe that the typical diagnostic features of subtrochanteric fractures are atypical femoral fracture pattern associated with bisphosphonate use and very narrow IM canal.

In conclusion, although time to bony union is much longer in pycnodysostosis patients than in normal patients, fracture union after extramedullary osteosynthesis can be obtained through either primary or secondary bone healing, and secondary bone healing might be advantageous in terms of fracture healing time.

Author Contributions All the authors contributed to the study conception and design. Material preparation, data collection and analysis were performed by all the authors. The first draft of the manuscript was written by the corresponding author and all the authors commented on previous versions of the manuscript. All the authors read and approved the final manuscript.

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Declarations

Conflict of Interest The authors declare that they have no conflict of interest.

Ethical Approval Local Ethics committee approval was taken for the management of this case.

Informed Consent Informed consent was not needed from all the individual participants included in the study.

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