Pathological fracture in fibrous dysplasia: a case report

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ABSTRACT

Fibrous dysplasia (FD) is described as a growth disorder characterized by the progressive replacement of normal bone elements by fibrous-osseous tissue. Bones affected by FD is presented with bone weakening and prone to pathological fracture. We reported a case of FD in an 8-years-old boy, who came to the hospital with pain in the upper right thigh after falling with bumps in his head and foot. An X-ray revealed a deformity as a diasteal scalloping with radiolucency lesions on one-third proximal of the right upper femur, greater trochanter, and lesser trochanter, with complete transverse discontinuity in the distal deformity. No similar lesion was found in the other bones. Bone structure and trabeculation in the deformity area has decreased and the cortex was thinning. On magnetic resonance imaging (MRI), bone size was enlarged, hypointense on T1WI and inhomogeneous hyperintense on T2WI. Fibrous dysplasia with a pathological fracture is a rare case. The appearance on the X-ray was diasteal scalloping with a ground-glass radiolucency represented with Shepherd’s crook deformity. This lesion was accompanied by a pathological fracture (fragility fracture) on distal lesion. Fibrous dysplasia which characterized by bone developmental anomaly gave an inhomogeneous hypointense on T1W1 and hyperintense on T2WI.

Keywords: fibrous dysplasia; ground glass lucent; pathological fractures; X-ray; MRI
INTRODUCTION

Fibrous dysplasia (FD) is a congenital skeletal disorder which can be associated with benign tumor. Lichtenstein first described FD in 1938 as a growth disorder characterized by the progressive replacement of normal bone elements by fibrous tissue.\(^1\) Fibrous dysplasia is a developmental anomaly in which normal bone marrow is replaced by fibro-osseous tissue. This process can be localized to a single bone, a small segment, or affect the bone structure in a diffuse pattern.\(^2\) Bones affected by FD present with bone weakening, making it abnormally fragile and prone to fracture due to minor trauma.\(^4\) Muthusamy et al.\(^5\) reported that the incidence of FD is 5 - 10% among benign tumors. Fibrous dysplasia divided into some types with prevalence of each type as follow, monostotic-type fibrous dysplasia (70 - 80%), polyostotic fibrous dysplasia (20 - 30%), McCune-Albright syndrome (2 - 3%), and Mazabraud's syndrome (very rare cases).\(^5\) Fibrous dysplasia which is a common benign skeletal lesion, may involve one bone (monostotic) or multiple bones (polyostotic), and affect throughout the skeleton with a predilection for long bones, ribs, and craniofacial bones.\(^9\)

Fibrous dysplasia is a congenital disorder characterized by bone marrow replacement with soft tissue. Monostatic fibrous dysplasia patients with rapid bone growth had a higher chance of malignant transformation. It is important to make malignant bone tumor as differential diagnosis of FD, such as osteosarcoma, osteoblastomas, or metastatic bone lesion. On the other hand, FD carries a very small risk to be sarcomatous transformation (1%).\(^7\)

In radio-imaging, FD can be described as pagetoid, sclerotic, and cyst like appearance.\(^8\)

Fractures caused by FD are more frequent in childhood, between the age of 6-10 years and declining thereafter.\(^9\) Pathological fractures can occur in more than fifty percent of the patients with polyostotic and monostotic FD.\(^10\) Approximately, 5% of fibrous dysplasia were found in benign bone lesion, and monostatic form eight to ten more common than polyostotic.\(^12\)

The etiology of FD is linked to the developmental failure of immature bone and irregular bone tissue leading to the formation of woven bone mass in abnormal fibrous tissue.\(^10\) The defect is associated with a gene mutation encoding the subunit of the stimulatory G protein (\(G\_{\alpha}\)) located at 20q13.2-13.3.\(^6\) It was demonstrated that active mutation of \(G\_{\alpha}\) in osteoblastic cells of patients with McCune-Albright syndrome and monostotic-type disease leads to constitutive activation of adenylate cyclase, increased cell proliferation, and inappropriate cell differentiation, resulting in overproduction of disorganized fibrotic bone matrix.\(^10\)

Long bone fracture is the most common complication in FD. Proximal femur is the most common site of fracture which mainly caused by load transfer through weakened bone and repetitive microfracture that produce progressive varus and bowing known as Sherperd’s crook deformity.\(^9\) A coexisting aneurysmal bone cyst is a risk factor of pathologic fracture that causes bone weakened.\(^13\)

Radiological examinations to diagnose FD are conventional X-ray, CT scan, and MRI. The common radiographic features in FD were radiolucent images,
as a picture of a decrease in the matrix of bone material, resulting in a reduction in the bone structure and strength. Fractures triggered by trauma can be the major risk for patients with expansion of cortical thinning and bowed shape of the bone. We presented a case report of FD in pediatric with specific features on X-ray and magnetic resonance imaging (MRI).

**CASE**

An 8-years-old boy came to the hospital with a chief complaint of pain in the upper right thigh after a fall with bumps on his head and foot. He was slipped and fell on his right foot. He was unable to walk properly. The upper thigh was swollen, and radiographs were obtained.

Conventional X-rays showed deformity with bone enlargement and ground glass lucency lesions on the one-third proximal of the right femur, greater trochanter, and lesser trochanter, accompanied by complete discontinuity with an irregular fracture line that crossed the distal deformity area with a callus formation. The ground-glass lucency lesion was well-demarcated, smooth edges, and sclerotic thickened layer (rind sign), and no soft tissue involvement was seen. The bone structure and trabeculation around the deformity area showed thinning of the bone cortex. The epiphyseal line was still evident without any abnormalities were seen in the femoral head or acetabulum. The lesion showed shepherd’s crook deformity (FIGURE 1). The similar lesion was not found in the other bones.

![Figure 1](image1.png)

**FIGURE 1.** A. Lower limbs X-ray (anteroposterior view). B. Pelvic X-ray (anteroposterior view) A ground-glass expansive radiolucent lesion one-third proximal of the right femur gives a shepherd’s crook deformity with a fracture in the distal lesion (a). Soft tissue was normal (b).

The MRI showed a widening bone lesion on T1WI with hypointense in the one-third proximal of the right femur, greater trochanter, and lesser trochanter, accompanied by signs of fracture in the distal lesion with hyperintensity at the edges of the fracture line and callus. On T2WI, the MRI shows an increase in the intensity of the inhomogeneous signal.
In the image above, MRI showed the coronal section of the hypointense lesion on T1WI and inhomogeneous hyperintense on T2WI (FIGURE 2A and B), sagittal section (FIGURE 2C and D), accompanied by a pathological fracture in the distal lesion followed by a callus. Younger age patients with femoral lesions tend to have high potential mechanical deficit leading to fracture. The femoral bone weakened by FD was prone to fracture or deformity caused by high mechanical forces.

**DISCUSSION**

Fibrous dysplasia is a benign disorder that affects bone growth as marked with the replacement of bone with fibrous tissue. Radiographically, FD was depicted as a radiolucent area that develops into the ground glass. The typical features of the disease are endosteal scalloping, bone expansion, and a sclerotic thickening reaction called the rind sign. The MRI was more accurate for the detection and assessment of affected area by FD and was useful to determine a doubtful radiographic result of suspected FD. Our case exhibited similar features with case reported by Hakim et al., which presented radiolucency, endosteal scalloping, expansion, and the presence of a rind sign as the radiographic findings. In this case, we found a shepherd’s crook deformity and a rind sign in the upper femur area, which is typical for FD. In MRI finding, FD presented as hypointense on T1WI and can be either hyperintense or hypointense on T2WI. The signal intensity on T1 and T2WI images and the degree of contrast enhancement on T1 images depend on bone trabeculae, cellularity, collagen, and cystic and hemorrhagic changes.

The cystic changes in MRI were not found in our case, this condition was similar with previously case reported by Jee et al. Among 13 MRI of FD cases, cystic changes were observed in two samples (15%). The imaging similarity was found between giant cell tumor and cystic fibrous dysplasia in the long bone; hence the understanding of the difference was required to avoid misdiagnosis. Cystic FD can be proven by biopsy.
FIGURE 3. The pre surgery X-ray showed a pathological fracture and an expansive lucent lesion surrounded by a sclerotic lesion.\textsuperscript{6}

FIGURE 4. A. X-ray radiographs with radiolucent lesions with a well-defined sclerotic rim, B. T1WI, hypointense lesion, C. T2WI, hyperintense lesion.\textsuperscript{14}

In this case, a complete pathological fracture with callus remodeling was discovered in the distal lesion (FIGURE 1). In certain conditions, fractures in FD patients are not associated with the bone condition caused by FD itself. Decreased bone mineral in FD should be considered as risk factor for fracture.\textsuperscript{6,10,15,17} Pathological fractures should be suspected in pediatric patients with fracture-associated minor trauma, unusual fracture site, or abnormal bone process found on radiographs. Changes in normal bone biomechanics could be caused by intrinsic processes including changes in bone mineral density caused by bone tumors (both benign and malignant), illnesses such as osteogenesis imperfecta or infections, and extrinsic mechanisms including internal fixation, biopsy canal, and radiation. Load and changes in bone intensity both influence the risk of pathologic fracture.\textsuperscript{18}

Pathological fracture is often associated with pain and deformity, which was divided into micro or macro fractures. Microfractures commonly occur in the trabecular bone of the metaphysis or corpus vertebrae, generally this condition is undetected and mostly immobile.\textsuperscript{18} Characteristics changes in X-ray including pathognomonic ground-glass aspect with peripheral sclerotic reaction (rind sign), bone expansion, an indentation of areas inside the cortex (endosteal scalloping), and involved the femur represented the classical Shepherd’s staff deformity as the result of repetitive microfractures.\textsuperscript{18-20}
FIGURE 5. In a 14-year-old male patient with sudden onset of right hip pain, radiographs A and B at presentation show Shepherd deformity and pathological fracture of the proximal femur.\textsuperscript{18}

The pathological macrofracture in this case was seen on a distal lesion, whereas the microfracture was seen in the lesser trochanter and column of the femur forming shepherd's crook deformity (FIGURE 1). \textcite{de Mattos et al.\textsuperscript{18}} reported that pain, size of the lesion (>2.5 cm in width or >3.5 cm in length) and cortical destruction ($\geq$ 50%) are not independently predicted the risk factors of fracture. Further study is required to determine the risk factor of pathologic fracture.

CONCLUSION

Fibrous dysplasia with a pathological fracture, in this case, is a rare case. On conventional X-ray radiographs, the lesion showed endosteal scalloping with a ground-glass radiolucency, and shepherd's crook deformity, occurring on the right upper femur in an 8-years-old boy. The lesion is accompanied by a rind sign, which defined as the presence of a sclerotic area around the lesion. In this case, the lesion is accompanied by a pathological fracture (fragility fracture). Fibrous dysplasia gives an inhomogeneous hypointense on T1WI and hyperintense on T2WI. Hyperintense signal inhomogeneity on T2WI image indicates an internal condition of thinning bone within the cortex of the disease. It is important to recognize FD radiological findings because of the higher chance of malignant transformation. Therefore, biopsy is needed to exclude any malignant bone tumor.

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