

Fibrous Dysplasia with Massive Cartilaginous Differentiation (Fibrocartilaginous Dysplasia) in the Proximal Femur: A Case Report and Review of the Literature

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Key Words

Fibrous dysplasia · Cartilaginous differentiation · Fibrocartilaginous dysplasia · Proximal femur

Abstract

Fibrous dysplasia (FD) is a monostotic or polyostotic benign bone lesion with spindle-cell proliferation in woven bone and stroma. Rarely, cartilaginous differentiation can be seen in the lesions of FD. FD with massive cartilaginous differentiation is called fibrocartilaginous dysplasia (FCD) and is considered a rare variant of FD. Although pathological findings of FD show irregular immature bone formation without osteoblastic rimming in fibrous tissue, and rarely show very small amounts of cartilage, histological images of FCD are said to show that cartilage with a relatively high cell density is present in the majority and that FD-like findings are seen in parts of it. The most characteristic feature of FCD on images is calcification in the lesions reflecting cartilaginous tissue. On the other hand, typical radiographic findings of FD include shadows with a ground-glass appearance and thinning and bulging of the cortical bone, the observation if calcification is not usual. Therefore, in the diagnosis of FCD, differentiation from multiple enchondromatosis, Ollier disease, chondrosarcoma, and chondrosarcoma secondary to FD is necessary, and it seems important to make a careful diagnosis based not only on the pathological findings but also on imaging and clinical findings. Herein, we report on a case of FD of the proximal femur associated with intralesional extensive carti-

laginous differentiation in which a pathological fracture occurred during follow-up, with a review of the literature.

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Published by S. Karger AG, Basel

Introduction

Fibrous dysplasia (FD) is a monostotic or polyostotic benign bone lesion with spindle-cell proliferation in woven bone and stroma [1]. Polyostotic FD is sometimes associated with McCune-Albright syndrome. Both monostotic and polyostotic FD are associated with missense mutations in the *GNAS* (guanine nucleotide-binding protein/ α -subunit) gene, which encodes the G-protein α -subunit involved in signal transduction via membrane receptors [2].

Histopathological images show characteristically shaped (C-shaped) woven bone and, in typical cases, bone formation without osteoblastic rimming on the background of fibrous proliferation containing collagen fibers. Although very rare, cartilaginous differentiation is seen in the lesions of polyostotic FD or FD of the proximal femur [3]. FD with massive cartilaginous differentiation is also called fibrocartilaginous dysplasia (FCD) and is considered a rare variant of FD [4]. Herein, we report on a case of FD of the proximal femur associated with intralesional extensive cartilaginous differentiation in which a pathological fracture occurred during follow-up, with a review of the literature.

Case Report

An 11-year-old boy with pain in the right hip starting in April 2014 presented to a local hospital. Plain radiography showed an abnormal shadow in the right proximal femur, and he was referred to our department for further examination and treatment. Local findings at the initial examination showed pain upon pressure and tapping tenderness on the outside of the right proximal thigh. His biochemical blood tests showed no obvious abnormal values except an increase in alkaline phosphatase (ALP) to 920 IU/l. Plain radiography revealed a bone lesion with a ground-glass appearance associated with a mild increase in the transverse diameter of the bone and thinning of the cortical bone from the metaphysis to the diaphysis of the right proximal femur. A marked calcification-like shadow associated with a pathological fracture without displacement was seen inside the lesion (fig. 1a). In addition, multiple areas of bone thickening of the skull with a ground-glass appearance were observed (fig. 1b). The lesion showed isointensity with punctate low signal intensity in some areas on T1-weighted magnetic resonance imaging (MRI) images (fig. 2a) and irregular high-signal intensity on T2-weighted images (fig. 2b). Irregular peripheral predominant enhancement was seen on gadolinium (Gd) contrast-enhanced images (fig. 2c). Bone scintigraphy revealed an accumulation of radioactivity in the right ilium, right proximal femur, right proximal ulna, and skull (fig. 3). The above imaging findings led to the diagnosis of pathological fracture associated with a bone tumor in the right proximal femur. Polyostotic FD and multiple enchondromatosis were considered as differential diagnoses.

Conservative treatment with no weight bearing on the affected limb was performed immediately, and bone union was achieved at the site of the pathological fracture during the course of treatment. After that, the lesion did not show any trend toward enlargement or other changes on images, and the subjective symptoms such as pain resolved. Therefore, the patient was kept under observation.

In November 2014, the patient again felt pain in his right hip when he performed a running long jump in a physical education class, and he presented to our emergency room. Plain radiography showed a pathological fracture with displacement in the metaphysis of the right femur (fig. 4a). Surgery was performed. During the surgery, artificial bone was implanted after curettage and fixed with a locking plate so that the varus of the proximal femur was corrected (fig. 4b).

Histopathological findings of the specimen obtained by curettage showed that nodular hyaline cartilage tissue was present in the majority of the specimen without marked atypia (fig. 5a). Proliferation of fibroblast-like spindle cells and woven bone were evident in part of the lesion (fig. 5b), and his final diagnosis was FCD. Two months after surgery, bone union was seen at the site of the pathological fracture, and the patient has had a good clinical course so far.

Discussion

FCD is a disease that was reported for the first time by Pelzmann et al. in 1980 as an FD lesion containing extensive cartilage and considered to be a subtype of FD. FCD can be said to be very rare, with only 13 cases reported in English so far (table 1). Histopathological images show extensive cartilaginous tissue in the lesion and, in some cases, chondrocytes with an increased cell density or atypia. Therefore, differentiation from bone tumors with similar histological findings, such as enchondroma or chondrosarcoma secondary to FD, is sometimes required.

We conducted a review of the 14 cases of FCD that have been reported so far, including our current case (table 1). According to these reports [5–11], there are slightly more males affected, the age of onset ranged widely from 4 to 53 years (average 18.8 years), the proximal femur (the site of the FCD in our case) was the most common site, and the ratio of polyostotic to monostotic disease was approximately 1:3. The most characteristic feature of FCD on images was calcification in the lesions reflecting cartilaginous tissue. On the other hand, typical radiographic findings of FD include shadows with a ground-glass appearance and thinning and bulging of the cortical bone, the observation of calcification is not usual.

In the present case, plain radiography revealed very similar findings to FCD. Although pathological findings of FD show irregular immature bone formation without osteoblastic rimming in fibrous tissue, and rarely show very small amounts of cartilage, histological images of FCD are said to show that cartilage with a relatively high cell density is present in the majority and that FD-like findings are seen in parts of it.

Therefore, in the diagnosis of FCD, differentiation from multiple enchondromatosis, Ollier disease, chondrosarcoma, and chondrosarcoma secondary to FD is necessary, and it seems important to make a careful diagnosis based not only on the pathological findings but also on imaging and clinical findings.

Statement of Ethics

The authors have no ethical conflicts to disclose. Informed consent was obtained from the patient and parents for this case report and any accompanying images.

Disclosure Statement

The authors have no conflict of interest directly relevant to the content of this article.

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Table 1. Reported cases of FCD

Case No.	Age, years	Sex	Location	Type of FD	Symptoms	Calcification on plain radiograph	Treatment	Reference
1	20	M	Proximal femur	Polyostotic	Mass	+	Osteotomy	[5]
2	23	F	Proximal tibia	Polyostotic	NS	+	NS	[6]
3	8	F	Proximal femur	Monostotic	Pain	+	NS	[6]
4	20	M	Proximal femur	Monostotic	Pain	-	NS	[6]
5	26	F	Ischiopubic bone	Monostotic	Mass	+	NS	[6]
6	14	F	Proximal femur	Monostotic	NS	+	NS	[6]
7	25	M	Proximal femur	Monostotic	Pain, fracture	+	NS	[6]
8	4	M	Femoral shaft	Monostotic	Limp	-	NS	[6]
9	15	M	Proximal femur	Polyostotic	Pain	+	Resection	[7]
10	53	M	Proximal femur	Monostotic	Pain	+	NS	[8]
11	21	M	Proximal femur	Monostotic	Progressive deformity	+	Amputation	[9]
12	6	F	Proximal femur	Monostotic	Pain	+	Curettage	[10]
13	17	F	Proximal femur	Monostotic	Pain	+	Resection of bone graft (homo)	[11]
14	11	M	Proximal femur	Polyostotic	Pain, fracture	+	Curettage of bone graft (artificial)	Current case

NS = Not specified.



Fig. 1. Plain radiography. **a** A ground-glass appearance with marked calcification and a pathological minor fracture in the right proximal femur. **b** Multiple ground-glass appearances in the skull.

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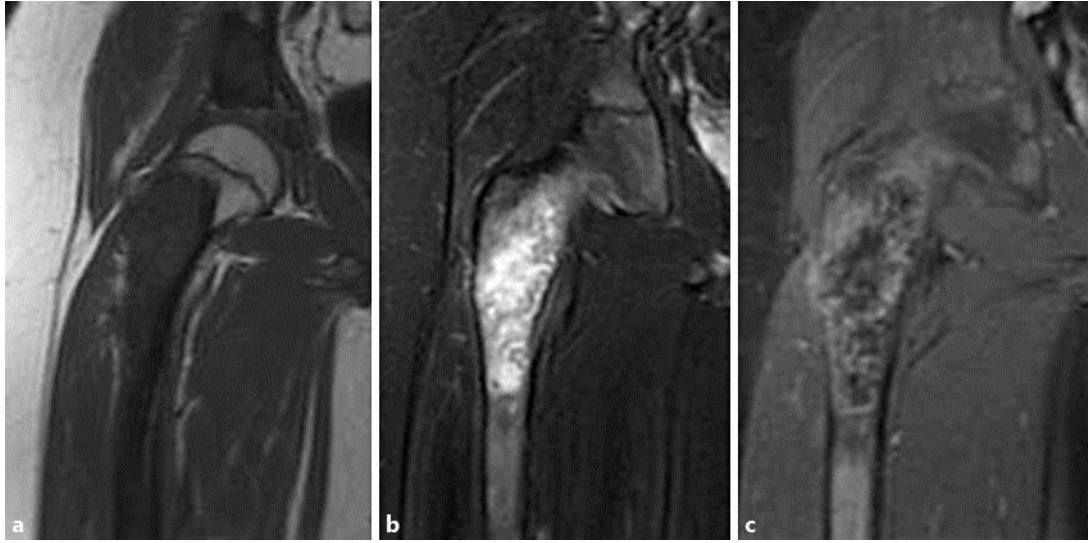


Fig. 2. MRI. **a** Isointensity with punctate low signal intensity on T1-weighted images. **b** Irregular high signal intensity on T2-weighted images. **c** Irregular peripheral predominant enhancement on Gd contrast-enhanced images.

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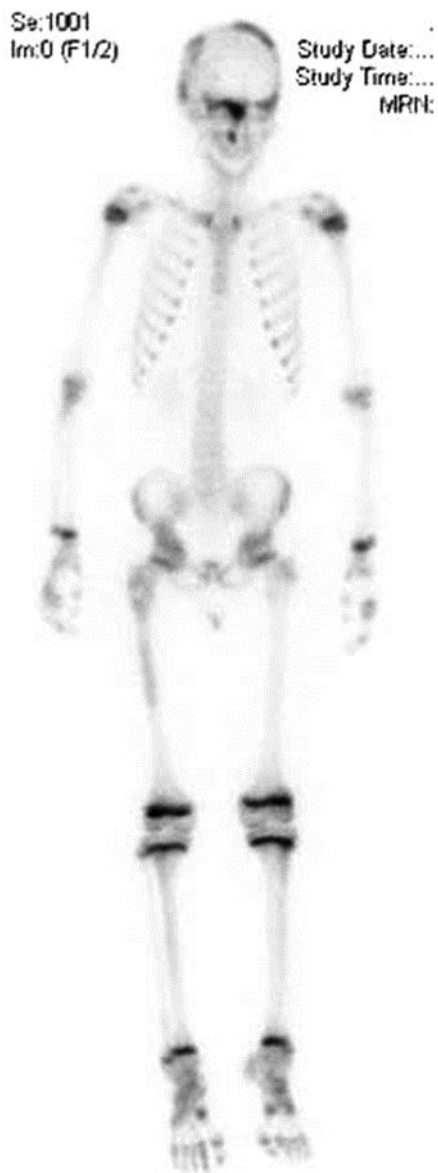


Fig. 3. Bone scintigraphy showing an accumulation of radioactivity in the right ilium, right proximal femur, right proximal ulna, and the skull.

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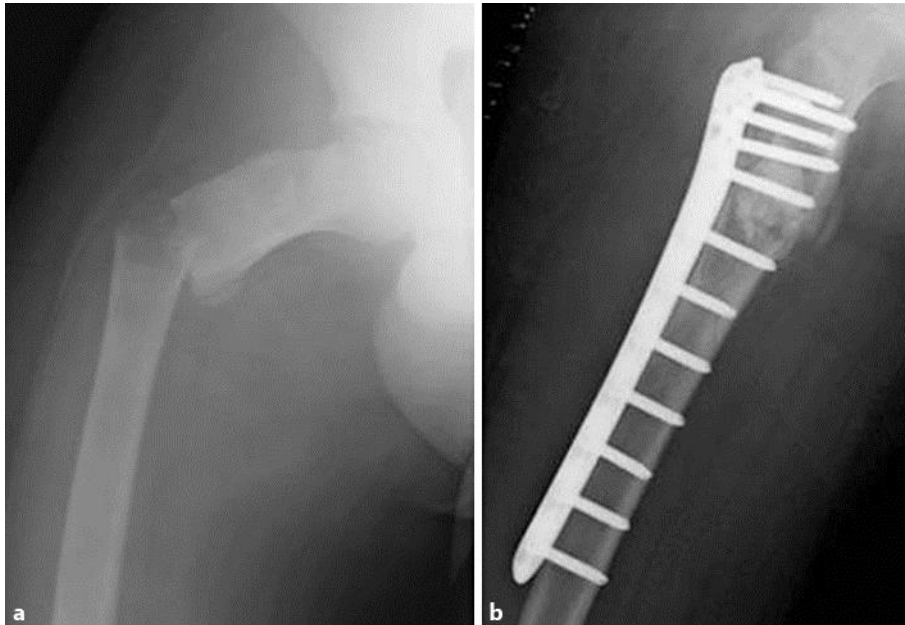


Fig. 4. **a** Pathological fracture with displacement in the metaphysis of the right femur. **b** Artificial bone was implanted after curettage and fixed with a locking plate so that the varus of the proximal femur was corrected.

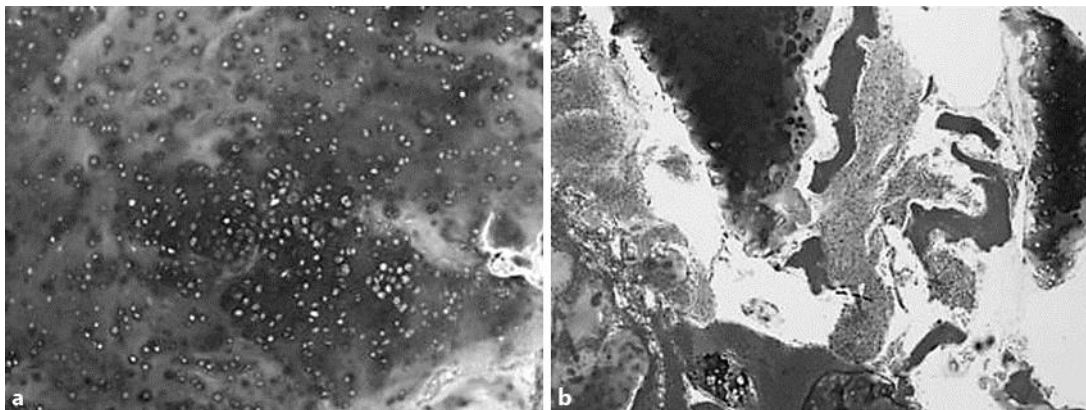


Fig. 5. Histopathological findings of the specimen. **a** Nodular hyaline cartilage tissue is present in the majority of the specimen without marked atypia. **b** Proliferation of fibroblast-like spindle cells and woven bone were evident in parts of the lesion.