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Fibrocartilaginous dysplasia (fibrous dysplasia and massive cartilaginous differentiation): Case report and literature review

Fatih Yılmaz [1], Funda Canaz [1], Evrim Yılmaz [1], Ulukan İnan [2]

[1] Department of Pathology, Eskişehir Osmangazi University, Faculty of Medicine Hospital, Eskisehir, Turkey
[2] Department of Orthopedics and Traumatology, Eskişehir Osmangazi University, Faculty of Medicine Hospital, Eskisehir, Turkey

ORCID ID of the author(s)
FY: 0000-0001-8216-1753
FC: 0000-0002-5642-3876
EY: 0000-0003-1937-8313
Uİ: 0000-0002-1903-5516

Corresponding Author
Fatih Yilmaz
Eskişehir Osmangazi University, Faculty of Medicine Hospital, Department of Pathology, Eskisehir, Turkey
E-mail: faatiihyiilmaaz@yahoo.com.tr

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Abstract

Fibrocartilaginous dysplasia (fibrous dysplasia and massive cartilaginous differentiation or fibrochondroplasia, FCD) is a rare variant of fibrous dysplasia and a term used for cases of fibrous dysplasia with prominent cartilage tissue. A limited number of FCD cases have been reported in the literature, which can be seen in both clinical forms.

A 16-year-old male patient, who had been followed for ten years with a diagnosis of polyostotic fibrous dysplasia in the left hip and cranium, presented with pain in the left leg after a fall. A subtrochanteric pathological fracture in the left femur was detected on exam, the lesion area was curetted, and osteotomy and fixation were applied. Microscopic assessment revealed a fibro-osseous lesion of benign spindle cell fibrous connective tissue with woven bone trabeculae, without osteoblastic rim or large areas of benign cartilage nodules. The final diagnosis was fibrocartilaginous dysplasia. In our literature review, 26 cases of FCD were reported so far. Age distribution of patients ranged from 4 to 53 years (mean 15.9) and the male / female ratio was 15/11 = 1.36. Eighteen cases were monostotic, and 8 were polyostotic. In cases with noted clinical and follow-up data, symptom duration ranged from 8 weeks to 18 years (mean 62.2 months), with no recurrence or malignant transformation in a mean follow-up of 21.71 (2-60) months post-treatment. In the cartilage component, there was increased cellularity, some nuclear atypia, binucleation, and myxoid degeneration. This situation simulates benign and malignant entities such as enchondroma, fibrocartilaginous mesenchymoma, well-differentiated intramedullary osteosarcoma, and chondrosarcoma with a differential diagnosis. FCD is a benign and very rare lesion with a prominent chondroid component, but may cause difficulty with differential diagnosis. Awareness of the histopathological and radiological features of FCD cases, their age range, and involvement areas provides an approach to distinguish them from lesions that may be confusing in a differential diagnosis.

Keywords: Fibrocartilaginous dysplasia, Fibrous dysplasia and massive fibrocartilaginous differentiation, Fibrous dysplasia, Fibrochondroplasia, Enchondral ossification, Chondrosarcoma

Introduction

Fibrous dysplasia (FD) is a dysplastic disorder of bone tissue characterized by woven bone structures that are randomly distributed, without an osteoblastic rim, within fibroblast-like spindle cell proliferation. It has been reported that GNAS1 gene mutations are found at a high rate in FD cases and play a role in their pathogenesis [1]. It has two clinical forms, monostotic and polyostotic. While the monostotic form is seen 8-10 times more frequently and may be asymptomatic, findings such as larger lesions, skin spots (café au lait), endocrine anomalies, and early puberty (McCune Albright Syndrome) are more common in the polyostotic form [2]. Although its exact prevalence is difficult to determine, given some asymptomatic cases, it accounts for approximately 5% to 7% of benign bone tumors [3].

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On microscopic examination, FD may present cementoid bodies, bleeding areas, secondary fibrohistiocytic proliferations, giant cell reactions, myxoid changes in stromal tissue, reactive bone areas of prominent osteoblastic rim, and cystic changes [1]. In addition, while microscopic cartilaginous differentiation can be found in approximately 10% of cases, cartilaginous matrix areas may be microscopically and radiologically dominant in rare cases [4, 5]. The terms "fibrous dysplasia and massive cartilaginous differentiation," "fibrocartilaginous dysplasia" (FCD), or "fibrochondroplasia" are suggested for these rare issues [1, 3, 6, 7]. We present a case who was followed for polyostotic fibrous dysplasia and operated on with a diagnosis of fibrocartilaginous dysplasia after a pathological fracture of the left proximal femur.

Case presentation

A 16-year-old male patient had calvarial thickening and ground-glass densities of the entire frontal bone as seen in a brain tomography 10 years ago; these findings are consistent with fibrous dysplasia. While he was followed with a diagnosis of polyostotic fibrous dysplasia in the left proximal femur and frontal bone, he was examined at our institution, with an operation plan a year ago. At that time, he had complaints of left hip pain and limping for 1 year. Physical exam revealed bilateral swelling on the frontal bone, and a café au lait spot with a size of 1 x 1 cm on the back. The radiological examination also revealed “Shepherd's crook” deformity of the left femur (Figure 1). Other physical findings were normal, with no signs of hyperthyroidism, hyperparathyroidism, hypercalcemia, or precocious puberty. The patient then presented with pain in the left leg after a fall. A subtrochanteric pathological fracture in the left femur was detected, so the lesion area was curetted, with osteotomy and fixation applied (Figure 2).

Figure 1: Antero-posterior radiograph of the left femur and classic shepherd's crook deformity

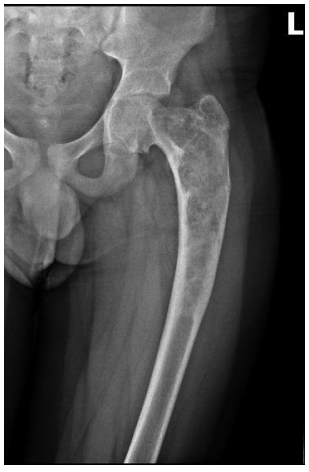
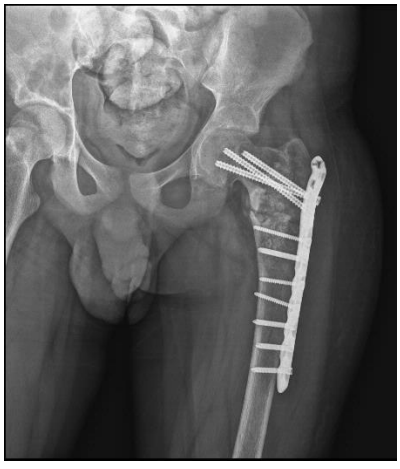


Figure 2: The radiological appearance of the lesional area after curettage, osteotomy, and fixation.



The patient’s operative specimen was macroscopically 7 x 7 x 4 cm in size, gray-brown, gray-white, and partly bone-hard. Microscopic examination revealed a fibro-osseous lesion of benign spindle cell fibrous connective tissue with woven bone trabeculae, without an osteoblastic rim (Figure 3) or large areas of benign cartilage nodules. The cartilaginous component was separated from areas of classical fibrous dysplasia with a distinct border, constituting 70% of the lesion (Figure 4). Hyperchromasia, atypia, increasing cellularity, and atypical mitosis were not observed either in the fibrous component or the cartilage nodules (Figure 5). The final diagnosis was fibrocartilaginous dysplasia, while informed consent was obtained from the patient’s family for scientific presentation.

Figure 3: Classical fibrous dysplasia area consisting of woven bone trabeculae (like Chinese letter) without osteoblastic rim within benign spindle cell fibrous connective tissue (H&E, 10X).

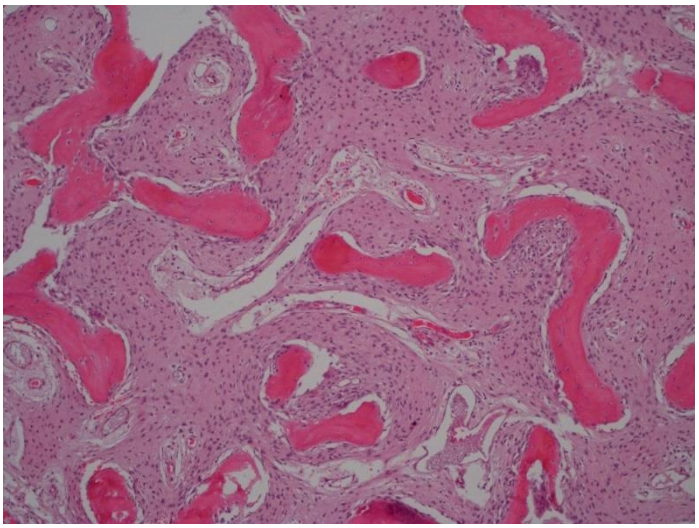


Figure 4: Classical fibrous dysplasia area and cartilage component separated by a distinct border (H&E, 4X).

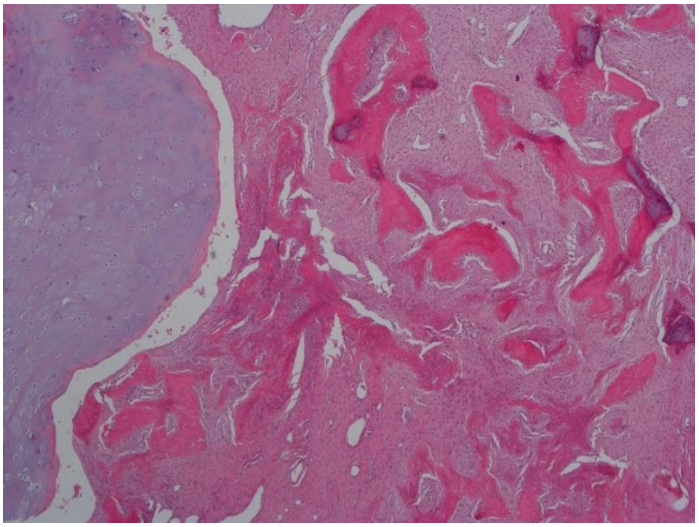


Figure 5: Enchondral ossification areas were observed around the cartilage nodules (H&E, 10X).

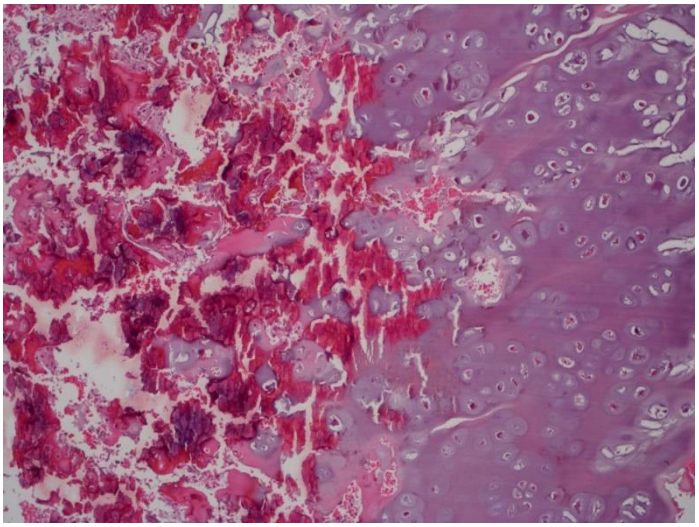


Table 1: Characteristics of reported FCD case

Case No. (reference)	Age	Gender	Localization	Type	Symptom	Symptom time	Follow-up
1. [7]	20	M	Proximal femur	P	Swelling	15 y	-
2. [5]	15	M	Femoral neck, tibia, fibula	P	Swelling, pain	7 y	-
3. [6]	23	F	Proximal tibia	P	-	-	-
4. [6]	8	F	Proximal femur	M	Pain	7 m	-
5. [6]	20	M	Proximal femur	M	Pain	-	
6. [6]	26	F	Ischiopubic bone	M	Swelling	8 y	
7. [6]	14	F	Proximal femur	M	-	-	
8. [6]	25	M	Proximal Femur	M	Pain, fracture	2 y	
9. [6]	4	M	Femoral shaft	M	Limping	-	
10. [4]	53	M	Proximal femur	M	Pain	10 m	-
11-18. [15]	8-18 (Mean: 11.3)	4 M 4 F	7 Femoral neck 1 Tibia diaphysis	8 M	Pain or fracture	-	-
19. [8]	21	M	Proximal femur, distal tibia	P	Pain, swelling, deformity	18 y	6 m
20. [14]	6	F	Femoral neck and proximal shaft	M	Pain	2 y	5 y
21. [13]	19	M	Proximal femur, distal fibula	P	Pain	3 m	1 y
22. [12]	8	F	Femoral neck	M	Pain, fracture	8 w	3 y
23. [11]	18	F	Femoral shaft, proximal radius and humerus	P	Pain	2 y	2 y
24. [9]	17	M	Proximal femur	M	Pain, swelling and progressive deformity	4 y	1 y
25. [10]	11	M	Proximal femur and skull	P	Pain, fracture	2 y	2 m
26. [17]	16	M	Right lower leg and foot	P	Tibial deformity, decreased mobility, and chronic pain	6 y	-
27. [Present case]	15	M	Proximal femur and skull	P	Pain, fracture	10 y	9 m

M: male, F: female, P: polyostotic, M: monostotic, y: year, m: month, w: week

Discussion

FCD is a rare variant of FD, and in our literature review, 26 cases have been reported so far (Table 1). This number is thought to be higher, due to the presence of incorrect or insufficiently described cases [8]. The age distribution of patients ranged from 4 to 53 (mean 15.9) and the male / female ratio was 15/11 = 1.36. Eighteen cases were monostotic, 8 were polyostotic, with the most common location of the lesions in the proximal femur (80%). The most frequent symptoms are pain, swelling, deformity, pathological fracture, and limping, all of which have been reported [3-15]. In cases with clinical and follow-up data, symptom duration ranged from 8 weeks to 18 years (mean 62.2 months), with no recurrence or malignant transformation observed in mean follow-up of 21.71 (2-60) months after treatment. Our case had similar characteristics to those reported in the literature, with no recurrence observed during the 9-month follow-up.

Focal fibrocartilaginous dysplasia (FFCD) cases in the literature may be confused with FCD, due to the similarity of names. FFCD is a rare benign disease of unknown etiology that tends to affect long bones in children. Histologically, it may exhibit a variety of patterns, from dense fibrous and tendon-like tissue to fibrocartilaginous tissue [16]. The presence of cartilage tissue is not an essential feature of FFCD diagnosis [16], with classical FD areas for diagnosis of FCD not found in reported FFCD cases.

Although FCD is considered a variant of FD, there are some differences and similarities. In addition, "Shepherd's crook" deformity on radiography for lesions of the proximal femur is known to be highly diagnostic of FD [2], but it should be noted that it may be seen secondary to metabolic, congenital, infectious, and traumatic conditions [9]. This finding is seen in many FCD cases in the literature [5, 7-9], and was present in our case. Unlike FD, the involvement of skull bones in FCD occurs less often, but was observed in one case [10]. In our work, calvarial thickening and ground-glass densities, which involve the entire frontal bone as detected in brain tomography 10 years ago, were accepted as compatible with FD.

A possible source of the cartilage component of FCD is thought to originate from cartilaginous rests near the growth plate, or callus tissue formed secondary to the fracture or coexisting enchondromatosis [4, 5]. However, no specific ratio or threshold value was determined. It was reported that cartilage areas in classical FD cases are usually smaller than 1 cm [6], using terms such as massive, extensive, prominent, dominant, large, and striking for the cartilage component in FCD [6, 8, 11-13, 15, 17]. However, in some cases, this ratio emphasizes that it constitutes the majority (60-85%) of the lesion [10, 11, 13, 14]. In our case, the cartilage component constituted approximately 70% of the lesion.

While classical FD areas constitute the fibrous component of FCD, we find in the cartilage component increased cellularity, some nuclear atypia, binucleation, or myxoid degeneration [6, 14]. This situation involves benign and malignant entities such as enchondroma, fibrocartilaginous mesenchyma, well-differentiated intramedullary osteosarcoma, and chondrosarcoma for a differential diagnosis [1, 3, 6, 14, 15]. The cartilage islands adjacent to the fibro-osseous component (typically FD) is the most important diagnostic clue to distinguish FCD from other cartilaginous neoplasms - such as enchondroma and chondrosarcoma [6]. Enchondral ossification areas, like the epiphyseal growth plate seen in the periphery of the cartilage islands, are generally not seen in conventional chondroid neoplasms [1, 6].

Fibrocartilaginous mesenchymoma (FCM) is a lesion easily confused with FCD clinically, radiologically, and histopathologically [8]. Although they were thought to be the same entities in the past, it is reported that they represent genetically different lesions [18]. FCM does not cause the gross distortion seen in FCD, can destroy the cortex and extend into the soft tissue, does not involve multiple bones, and spindle cells may demonstrate mild nuclear atypia and hyperchromasia as useful for a differential diagnosis with FCD [8]. Although bone production occurs in FCM, it is in the form of trabeculae formed by enchondral ossification at the periphery of cartilage masses surrounded by osteoblasts, unlike the characteristic woven bone trabeculae of FD [8].

The presence of coarse bony trabeculae, atypical nuclei, and mitosis versus the thin, branching woven bone trabeculae of FD, provides a guide in the differential diagnosis of FD versus low-grade intramedullary osteosarcoma [1]. Another method in differential diagnosis is to show GNAS1 gene mutation localized on chromosome 20. It was reported that the GNAS1 mutation is found in 50-70% of FD cases, playing a role in pathogenesis of FD [2], which supports the diagnosis of FCD [12]. In fibro-osseous lesions of the head and neck region, although there are few reported cases, GNAS1 mutations in FCM have not been demonstrated [18, 19]. Yet, it was reported recently that it can be found in 55% of parosteal osteosarcoma cases, but rarely in low-grade intramedullary osteosarcoma [19].

Treatment methods such as curettage of the lesion area, osteotomy, correction of deformity, and internal or external fixation are applied for the treatment of FCD [1, 12, 20]. Although a malignant transformation is rarely seen (less than 1%) in FD, there were none observed in any FCD case reported so far.

Conclusion

FCD is a rare benign lesion with a prominent chondroid component, so may cause difficulty with a differential diagnosis. The histopathological and radiological features of FCD cases, their age range, and involved areas should provide an accurate approach to distinguish them from lesions that may be confused with a differential diagnosis.

References

1. Czerniak B. Dorfman and Czerniak's Bone Tumors: Elsevier; 2016. 570-628 p.
2. Board WCoTE. Soft Tissue and Bone Tumours: International Agency for Research on Cancer, 2020. 472-4 p.
3. Dorfman HD. New knowledge of fibro-osseous lesions of bone. Int J Surg Pathol. 2010;18(3 Suppl):62-5.
4. Hermann G, Klein M, Abdelwahab I, Kenan S. Fibrocartilaginous dysplasia. Skeletal radiology. 1996;25(5):509-11.
5. Drolshagen LF, Reynolds W, Marcus N. Fibrocartilaginous dysplasia of bone. Radiology. 1985;156(1):32.
6. Ishida T, Dorfman HD. Massive chondroid differentiation in fibrous dysplasia of bone (fibrocartilaginous dysplasia). The American journal of surgical pathology. 1993;17(9):924-30.
7. Pelzmann KS, Nagel DZ, Salyer WR. Case report 114. Skeletal Radiology. 1980;5(2):116-8.
8. Kyriakos M, McDonald DJ, Sundaram M. Fibrous dysplasia with cartilaginous differentiation ("fibrocartilaginous dysplasia"): a review, with an illustrative case followed for 18 years. Skeletal radiology. 2004;33(1):51-62.
9. Vaishya R, Agarwal A, Gupta N, et al. Fibrocartilaginous Dysplasia of the Bone: A Rare Variant of Fibrous Dysplasia. Cureus. 2016 January 05;8(1):e448. doi: 10.7759/cureus.448
10. Morioka H, Kamata Y, Nishimoto K, Susa M, Kikuta K, Horiuchi K, et al. Fibrous dysplasia with massive cartilaginous differentiation (fibrocartilaginous dysplasia) in the proximal femur: a case report and review of the literature. Case reports in oncology. 2016;9(1):126-33.
11. Kaliyath S, Kishan Prasad H, Sajjan N, Mathias L, Shetty K, Rao C. Fibrocartilagenous dysplasia: A rare variant of fibrous dysplasia. Journal of Orthopaedics and Allied Sciences. 2015;3(1):37-9.
12. Henderson ER, Monforte H, Neustadt JB. Fibrocartilaginous Dysplasia of the Proximal Part of the Femur with a Pathological Fracture in a Child: A Case Report. JBJS Case Connector. 2014;4(4):e90.
13. Monappa, V., & Kudva, R. (2008). Multifocal fibrous dysplasia with enchondroma-like areas: Fibrocartilaginous dysplasia. The Internet Journal of Pathology, 7(2), 1-9.
14. Vargas-Gonzalez R, Sanchez-Sosa S. Fibrocartilaginous dysplasia (fibrous dysplasia with extensive cartilaginous differentiation). Pathology & Oncology Research. 2006;12(2):111-4.
15. Muezzinoglu B, Oztop F. Fibrocartilaginous dysplasia: a variant of fibrous dysplasia. Malaysian Journal of Pathology. 2001;23(1):35-40.
16. Bian Z, Lyu X, Guo Y, Zhu Z, Feng C, Yang J, et al. Focal fibrocartilaginous dysplasia of the distal femur. Journal of Pediatric Orthopaedics B. 2020;29(5):466-71.
17. Hakar MH, Chapman CM, Hansford BG, Davis JL. Polyostotic Fibrous Dysplasia With Extensive Cartilaginous Differentiation: A Rare Case Clinically Mimicking Ollier Disease. AJSP: Reviews & Reports. 2021;26(1):73-5.
18. Gambarotti M, Righi A, Vanel D, Cocchi S, Benini S, Elli FM, et al. Fibrocartilaginous mesenchymoma of bone: a single-institution experience with molecular investigations and a review of the literature. Histopathology. 2017;71(1):134-42.
19. Carter JM, Inwards CY, Jin L, Evers B, Wenger DE, Oliveira AM, et al. Activating GNAS mutations in parosteal osteosarcoma. Am J Surg Pathol. 2014;38(3):402-9.
20. Kishan Prasad H. Fibrocartilaginous dysplasia: A rare but distinct entity. Medical Journal of Dr DY Patil University. 2017;10(5):503-4.

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