Fibrous Dysplasia: Distinctive or Elusive? What to expect

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Learning objectives

To present a spectrum of varied, typical and atypical, imaging features of both monostotic and polyostotic form of Fibrous Dysplasia (FD), with special interest in the differential diagnosis.
Background

Introduction

FD is a rare, congenital, non-heritable entity, regarding benign intramedullary abnormalities either presenting as an isolated skeletal finding (monostotic type, M-FD), accounting for 70-80% of cases, or affecting multiple bones (polyostotic type, P-FD). P-FD can occasionally exhibit a monomelic pattern. The ribs are one of the most common sites of M-FD, while the femur and tibia are most commonly involved in P-FD. Craniofacial FD affects only the skull and facial bones, with the frontal, sphenoid, ethmoid, orbit, zygoma, maxilla, mandible, and temporal bones being the common sites. M-FD of the clivus is extremely rare.

FD can affect all age groups, most predominantly children and young adults, with no recognized gender predilection.

FD may manifest with a variety of clinical features, mostly depending on the age of onset, type of the disease, site(s) of involvement in the skeleton and associated endocrine disorders. It can often be asymptomatic, detected as an incidental finding on a radiological examination, particularly the monostotic type. When symptomatic, FD usually presents with pain, edema, bone deformities and/or fracture, causing morbidity. Clinical symptoms like recurrent sinusitis, epiphora, proptosis, central neurologic signs and neurovascular compression symptoms, can be seen in case of craniofacial FD.

P-FD may be associated with precocious puberty (McCune-Albright Syndrome/MAS), exostoses (fibrocartilaginous dysplasia) or intramuscular myxomas (Mazabraund’s syndrome).

The most frequent extra-skeletal manifestations are skin hyperpigmentation (café-au-lait spots) and precocious puberty, commonly seen in MAS.

A histologically similar condition, osteofibrous dysplasia (OFD), is characterized by fibro-osseous intracortical lesions extending to the medullary bone, almost exclusively occurring in the tibia or fibula of young children.

Imaging features

FD is usually diagnosed with plain X-rays, computed tomography (CT) and/or magnetic resonance imaging (MRI). Bone scintigraphy may be used to establish the extent and activity of the disease. MRI is the imaging study of choice in the follow-up process of young patients, owing to lack of radiation. Bone biopsy can be effective in case of unclear findings.
The classic radiographic appearance of FD is a well-circumscribed lesion, usually of ground-glass matrix but also radiolucent or mixed lytic/sclerotic, surrounded by a layer of sclerotic reactive bone (rind sign). This pattern results from defective mineralization of immature dysplastic bone. The classic rind sign is most commonly seen in the proximal femur. FD also causes expansion and remodeling of the involved bone, as well as bowing deformities associated with lesions in the long bones. A coxa varus angulation of the proximal femur is frequently seen in femoral involvement of FD (shepherd crook deformity). Shepherd crook deformity may also be seen in other disorders such as Paget disease of the bone and osteogenesis imperfecta. Typically, there is no periosteal reaction associated with FD lesions. Wide, transverse lucencies (looser zones) or true fractures may be seen in the deformed bone.

CT depicts FD lesions mostly as ground-glass opacities but they can also appear homogeneously sclerotic or mixed sclerotic/lytic, with well-defined borders and varying degrees of bony expansion and endosteal scalloping. CT scanning may be required to assess difficult regions such as the skull base, the spine and the pelvis. CT can better define the extent of the lesion and the presence of secondary aneurysmal bone cysts and is often used for quantifying the optic canal diameter in cases of craniofacial involvement.

On MRI, lesions usually appear as hypointense on T1-WI (isointense to muscle) while on T2-WI there is considerable variability in signal intensity (low, intermediate, high), mostly depending on the trabecular bone volume and the amount of collagen within the lesion. Contrast MRI typically shows delayed enhancement, with variable patterns (homogeneous, heterogeneous, central, ring-like). Contrast MRI is particularly useful for detecting cystic degeneration, aneurysmal bone cysts and soft-tissue involvement. It is also indispensable for the assessment of nervous structures in case of craniofacial FD.

Although FD is usually a straightforward radiographic diagnosis, on occasion it may be confused with malignant entities, because it has a rare potential of malignant transformation, with an increased frequency in the polyostotic form, especially in patients with Albright or Mazabraund's syndrome. Malignancies in M-FD are extremely rare. The most frequent entities to be considered in the differential diagnosis are central osteosarcoma and chondrosarcoma, especially in the case of excessive calcification and soft-tissue ossification. Other controversial situations include increase in the size and/or osteolytic areas of an FD lesion and the presence of an expansile lytic M-FD lesion. Although imaging diagnosis may be easier in cases of P-FD or typical M-FD forms, false-positive and false-negative diagnoses are not uncommon.

Treatment
Conservative therapy includes administration of analgesics, calcium and vitamin-D supplements, and treatment of associated endocrine abnormalities in case of MAS. Surgery (intramedullary nails, bone resection, osteotomy) is indicated in case pain does not respond to the conservative treatment, especially in extended bone damage and presence of fracture or increased fracture risk. In the event of hearing/vision loss or optic nerve compression, craniofacial surgery is the treatment of choice.
A retrospective review of 15 patients with FD and 1 patient with OFD was conducted. Radiographs were available in 11 patients, MRI in 5 patients and CT in 3 patients.

M-FD was found in 11 patients, involving the pelvic bones (3 patients), the humerus (2 patients), the femur (2 patients), the fascial bones (2 patients), the tibia (1 patient), and the ribs (1 patient) [Fig. 1, 2, 3, 4].

P-FD was found in 4 patients. In one of them, the ipsilateral femur and tibia were affected whereas in the remaining three, multiple long and small tubular bones, the skull and spine were involved. The spine is a quite uncommon site of P-FD, being involved in about 15% of the cases.

Among patients with P-FD, one suffered from MAS [Fig. 14, 15, 16], another had multiple coexisting exostoses [Fig. 10], while malignant transformation of a femoral site of P-FD was seen in a third patient [Fig. 9].

The variability of the FD appearance, in some cases required differentiation mostly from Paget's disease of the bone, due to common radiological characteristics, mostly in case of mixed regions of sclerosis and lucency. The key to the differential diagnosis in craniofacial FD is that the inner diploic table is typically spared (not displaced), in contrast with Paget's. Furthermore, Paget disease has a predilection for the skull vault and usually spares the facial skeleton. In the long bones, it usually begins as a subchondral area of lucency. In addition, the classical appearance in Paget disease is expanded bone with a coarsened trabecular pattern, with sclerotic changes occurring much later in the disease process. In most of our patients, the diagnosis of FD was based on the typical pattern of ground-glass lesion with surrounding sclerotic rim. Demographics and lesion location are also different in case of Paget's disease.

It can be intricate to distinguish a malignant lesion from malignant transformation of an FD lesion. In our case, imaging revealed no well-defined osteolytic lesion within the "ground-glass" matrix and only minimal periosteal reaction. The presence of extended ossification in the adjacent soft-tissue made the diagnosis unclear.

The most consistent radiographic feature of FD-related malignancies in patients who have not been subjected to surgery is an ill-defined, mineralized, osteolytic lesion within or near areas of "ground-glass" opacity, associated with cortical destruction. It is to be noted
that demonstration of mineralization is not a specific sign of osteosarcoma associated with FD, and definite diagnosis should be based only on histology.

FD was an incidental finding in all but three patients. The most common symptom among symptomatic patients was rapidly increasing pain at the site of involvement, while one presented with endocrine disorders due to MAS.

Radiographic appearance varied from lucent/ground glass, mostly in younger patients [examples in Fig. 1, 3, 13] to mixed/sclerotic [Fig. 2, 5, 9], with or without the presence of calcification. FD lesions caused Shepherd Crook’s deformity in long bones [Fig. 7] and bone expansion in all affected bones, most striking in cases of craniofacial FD [Fig. 8, 13]. Lesions surrounded by a layer of sclerotic reactive bone (rind sign), were indicative of fibrous dysplasia [Fig. 1, 3, 7, 16].

CT and MRI were more sensitive for fascial and pelvic FD but not satisfactory enough for lesions of long bones.
Fig. 1: Monostotic FD: Plain radiograph depicts a well-defined lesion of ground-glass matrix with a sclerotic rim (rind), within the proximal femur (red arrow).

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**Fig. 2:** Monostotic FD, upper limb x-ray: Diffusely abnormal irregular trabecular pattern with areas of mixed lytic/sclerotic appearance in the humeral neck and proximal metaphysis and more lucent lesions in the humeral shaft, causing bony expansion.

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**Fig. 3:** Monostotic FD: Plain radiograph depicts a well-circumscribed ground glass lesion of the right proximal tibia, surrounded by a thick sclerotic layer (rind), with minimal osseous expansion (red arrow).

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Fig. 4: Monostotic FD on MRI: a) Femoral metadiaphyseal intramedullary lesion, exhibiting low signal on T1-WI (similar to muscle), with associated bone expansion and bowing deformity (red arrow). No cortical destruction or periosteal elevation is present. b) Sagittal T2-WI image shows a heterogeneous, slightly hyperintense, signal of the fibrous dysplastic lesion.
Fig. 5: Polyostotic FD, monomelic pattern: Diffusely abnormal irregular trabecular pattern with areas of mixed lytic/sclerotic appearance and ground-glass opacity in the proximal femur, and well-defined lytic lesions in the ipsilateral femoral shaft (red outlines).

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**Fig. 6:** Polyostotic FD, plain radiographs: a/b) Diffuse areas of ground-glass opacity and soap-bubbly trabeculation in both humeral bones, with associated bony expansion and regions of endosteal scalloping, c/d): Bony expansion, remodeling and ground-glass matrix involving the 1st, 2nd and 3rd left metacarpal bones and 2nd - 3rd right metacarpal bones.

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Fig. 7: Polyostotic FD, plain radiographs (same patient as in Fig.6): a/b) Diffuse areas of ground-glass matrix and multiple mixed lytic/sclerotic lesions with associated bony expansion and endosteal scalloping, in both femoral and tibial bones and the right fibula, with bowing deformation of the latter (arrow, b). There are prominent radiolucent lesions of the left femur. Note the soft tissue ossification adjacent to the left proximal femur (arrow, a) that raises the question of malignant transformation. c) Well-circumscribed lesion of hazy ground-glass opacity with a sclerotic rind causing neck deformity and expansion of the right proximal femoral shaft (red arrow). Ill-defined mixed lytic/sclerotic lesion of the left proximal femur with a coxa varus angulation, known as "shepherd crook deformity" (red outline). There is also adjacent soft-tissue ossification. d): Bony expansion, remodeling and ground-glass opacity involving the 1st and 2nd metatarsal bones in both feet.
**Fig. 8:** Craniofacial FD, CT: Bone window at the level of skull base shows expanded clivus with ground glass density and cortical thinning (red asterisk).

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Fig. 9: Polyostotic FD, x-ray: A femoral site of polyostotic FD with a diffuse ill-defined pattern at the femoral neck and proximal metaphysis, and prominent sclerotic areas of the proximal shaft with cortical thickening. Note the subtle periosteal reaction (arrow) and ossified soft tissue component (outline), at the level of the greater trochanter. This is a case of malignant sarcomatous transformation in FD (Secondary osteosarcoma arising in fibrous dysplasia, as proven by histology).

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**Fig. 10:** FD in the tibia: Axial CT image demonstrates a well-marginated lytic FB lesion in the right tibia (asterisk), associated with multiple exostoses (arrows).

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Fig. 11: Polyostotic FD, lumbar spine x-ray: Well-demarcated radiolucent lesion within the L5 vertebral body (arrow), consistent with FD. There is no cortical erosion.

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**Fig. 12:** M-FD of the pelvis: CT coronal (a) and axial (d) images portray a clearly defined lytic lesion of the left ilium, at the level of the iliac tuberosity (arrows), with sclerotic borders, thin internal septations and associated cortical thinning adjacent to the ipsilateral sacroiliac joint. Corresponding MRI depicts the lesion as slightly hypointense on T1-WI (arrows in b,e) and hyperintense on STIR images (c,f) with hypointense linear septations. There is moderate bony expansion and deformation.

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**Fig. 13:** Craniofacial FD: Axial CT images demonstrate bony expansile lesions of ground glass matrix, with intact cortex, involving the right frontal bone (arrow, a), zygomatic
arch and inferior orbital rim (outlines, b/c). The appearance is typical for FD. Patient was asymptomatic and diagnosis was made incidentally during trauma work-up.

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Fig. 14: P-FD in the setting of MAS (1): Patient presented with clinical symptoms of precocious puberty and GH insufficiency. Axial brain CT images (a) reveal multiple expansile ground-glass lesions of the skull, involving the occipital and temporal bones, the clivus, sphenoid bone and zygoma (asterisks), with remodeling of the bone and areas of cortical thinning. Lesions are depicted as isointense to muscle, slightly inhomogeneous, on pre-contrast T1-WI images (b,c), showing moderate enhancement on post-contrast T1-WI images (d,e).

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Fig. 15: P-FD in the setting of MAS (2): Same patient as in Fig. 14 with multiple FD lesions throughout the skeleton. Femur x-ray (a) depicts a well-circumscribed lesion of ground glass opacity in the right proximal femur (asterisk), with bony expansion and minimal cortical thinning. The is suspicion of associated insufficiency fracture (arrow). Transverse diaphyseal fracture of the deformed bone was treated with external fixation, as demonstrated on post-surgical x-ray (b).

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**Fig. 16:** P-FD in the setting of MAS (3): Same patient as in Fig. 14-15 with multiple FD lesions throughout the skeleton. Upper limb x-ray (a) reveals well demarcated ground-glass expansile lesions in the radius and ulna (asterisks), with a subtle "rind sign" of the ulnar lesion. Note the transverse fracture associated with the deformed radius (arrow). Plain radiographs further depict lesions of ground-glass matrix in the ipsilateral tibia and fibula (asterisks, b), and in several metacarpal bones (c), with associated bony expansion and marked deformation. These are typical FD lesions.

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Conclusion

FD is usually asymptomatic and can appear as an incidental finding with typical radiographic features. Though benign as a rule, it can rarely display locally aggressive behavior, mimicking malignancy. Comorbidities are common in P-FD and should be excluded by the work-up process.
References


