

Fibrous Dysplasia: Recent Developments and Modern Management Alternatives

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Abstract: Fibrous dysplasia is a benign skeletal lesion that may present in a monostotic or polyostotic form. The later form can have café-au-lait lesions with associated endocrinopathy, otherwise known as McCune-Albright syndrome. Mutation of the GNAS gene is responsible for the development of fibrous dysplasia. Pain, limp, deformity, and fractures are the main presenting symptoms. While any bone might be affected, proximal femoral involvement is the most problematic from an orthopaedic standpoint. Although medical treatment to limit the disease burden is available for the polyostotic disease, the mainstay of treatment in symptomatic cases is surgery. This current concept review is aimed at a comprehensive review of the current literature and recent developments in our understanding of fibrous dysplasia and novel treatments, with a focus on proximal femur deformity. We also explore the role of state-of-the-art technologies, including 3D printing, in the modern management of fibrous dysplasia.

Key Concepts:

- Treatment of fibrous dysplasia lesions should be focused on providing mechanical stability and improving function, and not on eradicating the lesion.
- All bone grafts and substitutes are prone to resorption, and therefore, should be augmented with orthopaedic hardware.
- Intramedullary reconstructive devices with two proximal screws are the hardware of choice due to their load-sharing properties.

Introduction

Fibrous dysplasia (FD) is a benign tumor-like condition in which tissue of fibro-osseous origin replaces the normal bony structure. The first reports of the disease can be traced back to 1936,¹ with the polyostotic disease first being described by Lichtenstein.² Similar to other benign bone lesions of the human skeleton, FD can be

diagnosed incidentally or in association with certain symptoms. Therefore, the true incidence and prevalence are uncertain, but it is estimated to comprise 5-7% of all benign bone lesions.³ There is no gender predilection, and the most common sites of involvement are the femur, tibia, craniofacial bones,⁴ and ribs.⁵ The disease has two main presentations:

- 1) Monostotic FD: the most common type, with most cases being asymptomatic.
- 2) Polyostotic FD: which may or may not be part of a syndrome, including:
 - McCune-Albright syndrome (MAS): Polyostotic FD with café au lait spots and endocrine disorders, especially precocious puberty.
 - Mazabraud syndrome: Polyostotic FD with multiple soft-tissue myxomas.⁶

This paper is aimed at a comprehensive review of the current literature on fibrous dysplasia, with a focus on orthopaedic manifestations, particularly proximal femur deformity, and modern management alternatives.

Pathogenesis

Genetics

Mutations in the GNAS (guanine nucleotide-binding protein/ α -subunit) gene is responsible for the development of fibrous dysplasia. GNAS encodes $G_s\alpha$, a cAMP pathway-associated G-protein subunit. The primary role of $G_s\alpha$ is to couple G-protein receptors to adenylyl cyclase, which in turn promotes receptor-stimulated production of intracellular cAMP and a subsequent downstream of effects. The normal $G_s\alpha$, in its inactive state, forms a heterotrimer with $G_s\beta$ and $G_s\delta$ subunits, with GDP bound to its binding site. Exchange of GDP for GTP activates the $G_s\alpha$ subunit, which dissociates from the trimer and activates adenylyl cyclase to form cAMP from ATP. Loss-of-function mutations of the GNAS gene result in a constitutionally inactive $G_s\alpha$ and has been found in several disorders of the endocrine system (e.g., pseudohypoparathyroidism).⁷ Gain-of-function mutations on the other hand, lead to the constitutive activity and abnormally increased cAMP signaling and leads to the phenotype of fibrous dysplasia.⁸

Two missense mutations have been implicated in the majority of patients: Arg201 and Gln227,⁹ which are

tested when a targeted analysis of the GNAS gene is performed. While these mutations have been found to be highly specific to FD, the sensitivity is low.¹⁰ Currently, there is a 90% detection rate with modern next-generation sequencing (NGS) techniques.^{9, 11} It should be noted that as FD is the result of a postzygotic somatic mutation and is not inherited, prenatal testing is not indicated. Also, the risk of the disease in future pregnancies is similar to that of the general population.

Histopathology

The characteristic features in FD are abnormal fibro-osseous tissue with irregular, under-mineralized woven bone, and varying degrees of cellularity.¹² The classic 'Chinese writing' or 'alphabet soup' histologic pattern is predominantly observed in long bone lesions. In this form, the stroma is low in cellularity, with curvilinear, irregular trabeculae of woven bone arranged in a discontinuous pattern.¹³ Increased bone resorption by typical osteoclasts is another feature, which translates to the typical lytic lesion seen on the radiographs. The use of bisphosphonates in an attempt to reduce bone resorption was based on this finding.

Clinical Presentation and Natural History

Most monostotic lesions are asymptomatic and diagnosed incidentally on radiographs taken for unrelated symptoms. The lesions are more frequently diagnosed in the second or third decades of life.^{14, 15} The femur is the most common site of involvement. Patients with polyostotic FD usually present at an earlier age with pain, limp, deformity, or fractures.^{14, 16} Children with McCune-Albright syndrome (MAS) tend to be diagnosed the earliest, typically during the first decade of life. The classic presenting symptoms are non-orthopaedic, most commonly signs of precocious puberty (e.g., vaginal bleeding).⁶

Monostotic lesions may expand in size during skeletal growth but typically stabilize after maturity. Complications, including fractures and deformity, occur

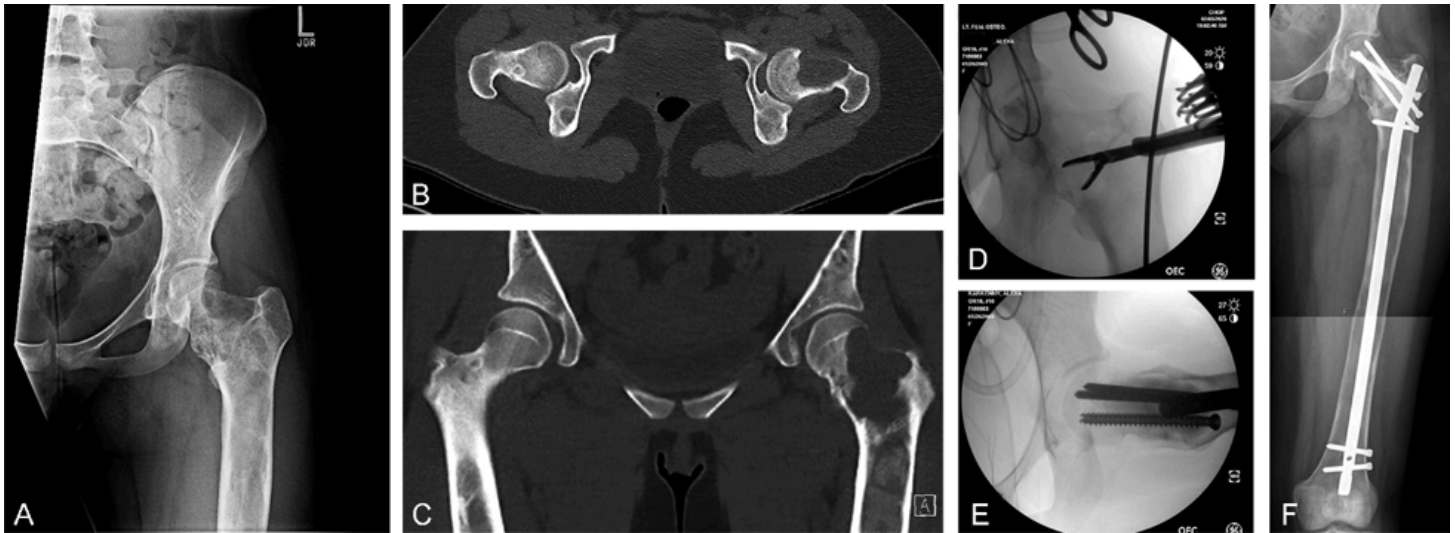


Figure 1. A 16-year-old female with McCune-Albright syndrome. A femur X-ray is suspicious for a cystic lesion (A). A CT scan of the pelvis shows cortical thinning and possible disruption with cystic changes (B, C). The patient was diagnosed with cystic degeneration and underwent surgery. A biopsy (D), confirmed the diagnosis of cystic degeneration. Fixation was achieved by a nail with two screws in the head and one 7.3mm freehand screw (E, F). Synthetic bone graft was also used.

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less frequently than with the polyostotic disease, and the long-term prognosis is favorable. Hip function is reasonably preserved in most cases, and despite mild leg length discrepancy (LLD), function tends to be minimally impaired.¹⁴ Han et al. reported a progression-free rate of 81% at 5-year follow-up in adults.¹⁷

Polyostotic disease (PFD) on the other hand, tends to be progressive beyond skeletal maturity. Ippolito et al. demonstrated femoral deformity progression in 87% of the cases, and a 41% rate of surgery required within a 7-year follow-up period.¹⁸ In PFD and MAS, long-term morbidity, including significant LLD, multiple fractures, chronic pain, and deformity are common. Poor functional outcomes are also significantly more common than with monostotic disease.¹⁴ MAS patients are more likely to require treatment, including medical (bisphosphonate therapy) and surgical treatment (e.g., stabilization of fractures, prophylactic intramedullary nailing). Scoliosis is another common occurrence in PFD with potentially devastating complications.

Associated Lesions and Malignant Transformation

Cystic degeneration of FD lesions was first reported by Simpson et al. in 1989.¹⁹ This locally aggressive form of the disease presents as a cystic lesion with cortical disruption and potential for soft tissue extension (Figure 1). Local and radicular pain may be present. While the clinical presentation and the radiographic findings suggest malignant transformation, histology shows typical FD.²⁰ These lesions may need to be treated with curettage and bone graft with or without prophylactic fixation due to a high risk of pathological fracture. Long-term follow-up of these lesions with serial radiographs is recommended.

Aneurysmal bone cysts (ABC) are often associated with other benign bone lesions. The association with fibrous dysplasia was first reported in 1971.²¹ There are only a few cases reported in the long bones of the extremities. There is no indication that the prognosis is altered by this association.^{22, 23}

Malignant degeneration may occur, but the frequency of such transformation is difficult to determine. The most common malignancies associated with FD are osteosarcoma, fibrosarcoma, and chondrosarcoma.³

Ruggieri et al. reported 28 cases of malignancies arising from FD lesions from the Mayo Clinic cohort.²⁴ Thirteen (46%) had a history of radiation therapy. Radiation therapy is now a well-known

potential carcinogenic for a myriad of benign bone lesions and is no longer used in the management of FD. Qu et al. reported 10 malignancies arising from monostotic FD without a history of radiation, including seven osteosarcomas, two fibrosarcomas, and one malignant fibrous histiocytoma (MFH).²⁵ It is extremely rare for a malignancy to be the presenting symptom of fibrous dysplasia. Therefore, any change in the pain levels without apparent trauma or FD progression should raise concerns for malignant transformation. Changes in the radiographic appearance of the lesion, including increased mineralization, expansion, and cortical destruction, should alert the clinician to evaluate the lesion further.

Radiographic Features

On plain radiographs, fibrous dysplasia presents as an intramedullary, well-defined, and mildly expansile lytic lesion.^{26, 27} The lesion most often involves the diaphysis and metaphysis, and the epiphysis seems to be spared.²⁸ The margins are well-defined, sclerotic borders may be present, and mild endosteal scalloping can occur (Figure 2-A).²⁶ There is no associated soft-tissue mass and

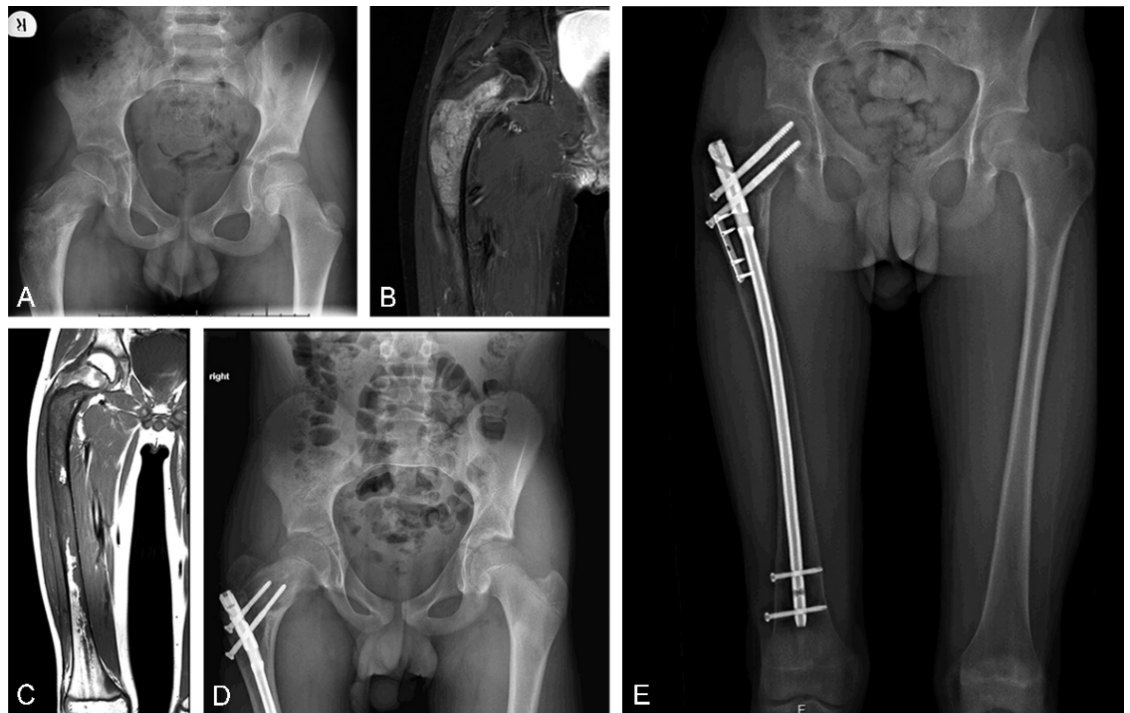


Figure 2. A 10-year-old boy with polyostotic fibrous dysplasia presented with a limp and a painful lesion in the right femur (A). MRI is characteristic, with intermediate signal on T2-weighted (B) and hypointense signal on T1-weighted images (C). Biopsy, curettage, and nailing was performed. Three years later, the patient had recurrent pain and limp, and coxa vara (D). Proximal femoral osteotomy and fixation with an intramedullary nail was performed. A locking plate with unicortical screws was used for provisional fixation during reaming and was left in place for added stability (E). He is pain-free at a 4-year follow-up.

periosteal reaction should be absent unless a pathological fracture is present.²⁸ The characteristic ‘ground-glass’ appearance is the result of a homogenous fibro-osseous matrix replacing the normal trabecular woven bone. Other radiographic presentations include pure osteoblastic lesions, which are rare, the presence of some trabeculae in the matrix, as well as islands of cartilage, which has been reported in up to 10% of patients.²⁸

CT scans demonstrate the extent of the disease, the degree of cortical thinning, and associated deformities

better than plain radiographs. CT is most valuable for preoperative planning of FD-related deformities, especially in the proximal femur. CT-based 3D modeling and patient-specific fixation devices are becoming more accessible. While CT scans better characterize the deformity, it is less useful for diagnosis than the MRI.³ MRI appearance of FD is very typical. There is a hypointense or intermediate signal on T1-weighted images, a hyperintense or intermediate signal on T2-weighted images, and heterogeneous enhancement on contrast-enhanced images.²⁹ MRI is also important in the differential diagnosis and to evaluate extra-osseous involvement in cases where malignant degeneration is suspected (Figure 2).

A skeletal survey is highly sensitive in determining the extent of skeletal involvement. Newer imaging technologies, however, such as low dose 2D/3D radiography (e.g., EOS), have become increasingly popular and have replaced the traditional surveys. At our institution, we routinely perform a low-dose lower extremity imaging for all newly identified fibrous dysplasia. We don't routinely scan upper extremity lesions, but that is a case-by-case decision.

Radionuclide bone scintigraphy shows non-specific increased tracer uptake and can be used to delineate the extent of skeletal involvement.³⁰ However, 'cold' scans are commonly encountered and bring the reliability of bone scan under question, especially given the amount of radiation and other modern techniques.³¹ FDG PET/CT (18 F-fluorodeoxyglucose positron emission tomography) scans are widely used to diagnose and stage malignancies and metastasis. Primary reports of PET/CT in FD were promising, but further studies showed a high false-positive rate, and multiple cases of FD mimicking malignancy/metastasis on PET/CT scans have been published.³² Therefore, not only is the role of PET in the evaluation of fibrous dysplasia controversial, but also a positive scan in a patient with a previously quiescent FD should be interpreted cautiously.³³

Treatment

Although most of the literature on fibrous dysplasia consists of case series with heterogeneous study populations, it is evident that the treatment and prognosis of fibrous dysplasia has improved with a better understanding of the disease pathophysiology. The shift from radiation therapy and nonsurgical management of fractures, towards adequate management of underlying endocrinopathies, and development of new surgical techniques and devices, have improved the prognosis and long-term outcome.

PFD and MAS are most effectively managed by a multidisciplinary team, including surgeons, endocrinologists, oncologists, clinical geneticists, and other specialists as needed. Best practice guidelines for the management of multiorgan involvement have recently been published, and offer a comprehensive tool in the hands of the clinicians.³⁴

Observation

Most upper extremities lesions, and monostotic lower extremity lesions with little minimal fracture risk, are amenable to nonoperative management. The frequency in which patients should be re-evaluated and lesions re-imaged depends on age, the extent of the lesion, and the presence of angular deformities or any symptoms. In general, our protocol is bi-annual follow-up visits with a thorough physical examination and radiographic surveillance for skeletally immature patients. For skeletally mature patients, regular radiographic follow-ups may not be necessary unless there are symptoms. However, the treating surgeon should have a low threshold for repeat imaging based on the patient's complaints, or when there is any suspicion of a complication (e.g., progressive deformity, malignant degeneration).

Pharmacologic Management

In polyostotic disease, pain is a common symptom, which ranges from intermittent and mild to chronic and

severe. Management with acetaminophen or NSAIDs are the first line of treatment and can be very successful.

Bisphosphonates decrease osteoclastic activity, thereby decreasing bone turnover. They have been highly successful in the treatment of osteoporosis and diseases of high bone turnover, including Paget disease of bone. The same mechanism was postulated to rein in the disease activity in fibrous dysplasia. However, after being used for 25 years in FD, there is still no consensus on the indications, dosage, and route of administration. Currently, the food and drug administration (FDA) considers FD an off-label indication for bisphosphonate treatment. Pain management and high bone turnover (as measured by serum biomarkers) in polyostotic fibrous dysplasia are the main indications for bisphosphonate treatment.³⁴ While bisphosphonates have been successful in controlling pain in polyostotic disease in children and adults, the results are still mixed,^{35,36} and it is not known whether fracture occurrence and deformity progression can be mitigated with the drug.

More recently, the receptor activator of nuclear factor- κ B ligand inhibitor *denosumab* has been evaluated in polyostotic FD patients with pain refractory to bisphosphonate treatment. The results have been promising, with half the patients reporting complete elimination of pain.³⁷ However, in addition to being an off-label use, until the results of clinical trials have been thoroughly assessed, denosumab should only be administered in centers with expertise in the treatment of fibrous dysplasia or other bone dysplasias. Treatment of specific endocrinopathies should be under the supervision of an endocrinologist familiar with fibrous dysplasia and is beyond the scope of this review.

Surgery

The main indications for surgery in fibrous dysplasia include recalcitrant bone pain, impending or pathologic fracture, progressive skeletal deformity, and concern for malignancy.

Some of the key factors to be considered:

- *Age and the extent of disease:* Monostotic or localized lesions are more amenable to observation as they don't tend to progress. Younger patients, especially children with polyostotic involvement, need to be more aggressively followed and treated, as deformity can be progressive.
- *Location:* Upper extremity lesions are more amenable to conservative treatment; however, there is still the risk of recurrent fractures. Majoor et al. reviewed 50 patients with humeral lesions and sought to determine the risk factors of fracture and indications for surgical intervention. They found that 54% of the lesions sustained at least one fracture. All the fractures were initially treated nonoperatively. However, eight patients eventually required at least one surgical intervention. An axial length >30mm, a circumferential cortical involvement >50%, and presence of cystic degeneration were associated with increased risk of fracture and might be considered as indications for prophylactic treatment of humeral lesions.³⁸

Lower extremity or weight-bearing areas such as the femur, are at a much higher risk of fractures and deformities and are less-than-ideal candidates for conservative treatment.

- *Associated deformity:* Progressive deformity is one of the main issues in FD, especially in the region of the proximal femur. Re-establishing the normal mechanical axis and maintaining the correction are the two main goals. Understanding the bone properties and residual growth potential is essential for a long-lasting surgical result.

The two main challenges from an orthopaedic standpoint are the biomechanical properties of the bone and the recurrence or persistence rate post-excision of the lesion. Due to the replacement of normal woven bone for a fibrous-osseous component, and cortical involvement

Type of Graft	Mechanical Stability	Rate of Resorption	Indications for Use
Cancellous (auto or allograft)	-	+++	Virtually none
Autograft cortical	++	++	Rare, reconstruction of defects, allograft unavailable
Autograft cortical vascularized	++	+	Reconstruction of large cortical defects (e.g., malignancy)
Allograft cortical	++	++	Strut in defects and femoral neck, morselized could be used instead of cancellous graft
Synthetic (CPC)	+	+	Provides immediate stability, virtually unlimited supply. Scarce data, but results are promising
Coral	-	+++	None

Table 1. Characteristics of bone grafts and bone graft substitutes and indications for use in fibrous dysplasia. CPC: tricalcium phosphate cement. ‘-’ lowest, ‘+++’ highest. (Adapted from DiCaprio MR, Enneking WF. *Fibrous dysplasia: pathophysiology, evaluation, and treatment.* JBJS. 2005 Aug 1;87(8):1848-64.)

present in the majority of the lesions, load-sharing intramedullary fixation methods are undeniably superior to other modalities, such as plates and screws, fixators, etc. Depending on the location and the bone size (canal capacity), hardware selection can be challenging.

While there are reports of deformity correction and limb lengthening with modern ring external fixators combined with elastic nailing,³⁹ external fixation is best avoided in fibrous dysplasia, especially in extensive disease. Conventional plates and screws have been historically used in fibrous dysplasia. The challenge is that non-locking plates act by converting insertional torque into compression forces. In the deficient fibro-osseous tissue of FD, this will only lead to screw pull-out and subsequent fixation failure. In localized or monostotic disease, plate and screws may be used to bridge the lesion and provide adequate mechanical stability.

The following principles should be followed when using plate and screw constructs in fibrous dysplasia: 1) Titanium alloy is preferred over stainless steel due to a more similar modulus of elasticity to normal bone.⁴⁰ 2) Locking plates and screws are recommended over

compression screws to act as an internal fixator. 3) The longest plate that spans the whole bone should be selected, which is particularly important in polyostotic disease. There are many reports of a small plate acting as a stress riser, leading to a fracture at the plate-bone junction. 4) In general, the maximum number of screws should be used. The mechanism of failure in locking plates is a simultaneous pull-out of the whole plate-screws construct. A higher number of screws adds to the total pull-out strength.

Intramedullary devices are the gold-standard for fixation in fibrous dysplasia because of their load-sharing properties. Elastic nails are generally reserved for the upper extremity, as they do not provide as stable a fixation as rigid nails. These are also used in the skeletally immature tibiae in which rigid nails may be contraindicated. Rigid locked nailing is the choice in other circumstances.⁴¹ There is a variety of commercially available devices to choose from in terms of diameter, length, metallurgy, and locking mechanism. The longest nail possible with a cephalomedullary configuration should be selected, and one or two fixation points in the femoral neck are ideal. In younger patients with a

narrow medullary diameter, humeral nails have been successfully used.⁴² In cases where a suitable intramedullary device is not available commercially, patient-specific devices could be ordered with custom specifications, including unconventional lengths, diameters, and locking mechanisms.⁴³

While fibrous dysplasia is not a *true* neoplastic process, the inherent “instructions” for bone replacement in the affected area do not disappear with the excision of the affected bone. Therefore, one should expect that after intralesional curettage of fibrous dysplasia, new fibrous dysplastic bone would re-grow/recur into that area, even when bone graft is used. There is definitely a difference in the rate of dysplastic transformation of different bone graft materials, and this should be considered during graft selection (Table 1). Depending on the type and volume of the graft, as well as the extent of the disease and age of the patient, the graft will, sooner or later, be resorbed into the dysplastic lesion. The highest risk is in younger patients with polyostotic disease in whom a form of cancellous graft is used. Enneking popularized utilizing allograft cortical strut grafts, which take more time to be resorbed.⁴⁴ Synthetic bone grafts (tricalcium phosphate) have also been used with some success. Alpha-tricalcium phosphate (calcium phosphate cement, CPC) provides immediate stability to the bone and may take the longest to be resorbed into the lesion.⁴⁵

Fixation alone has been successful in reducing pain and the risk of fracture,⁴⁶ but could be combined with curettage and grafting if deemed necessary. Isolated curettage and bone grafting has produced poor results, with frequent resorption of the graft, fractures, and the need for re-operation.^{47,48} Nevertheless, it could be considered in a monostotic lesion with a low risk of fracture, such as in upper extremity lesions.

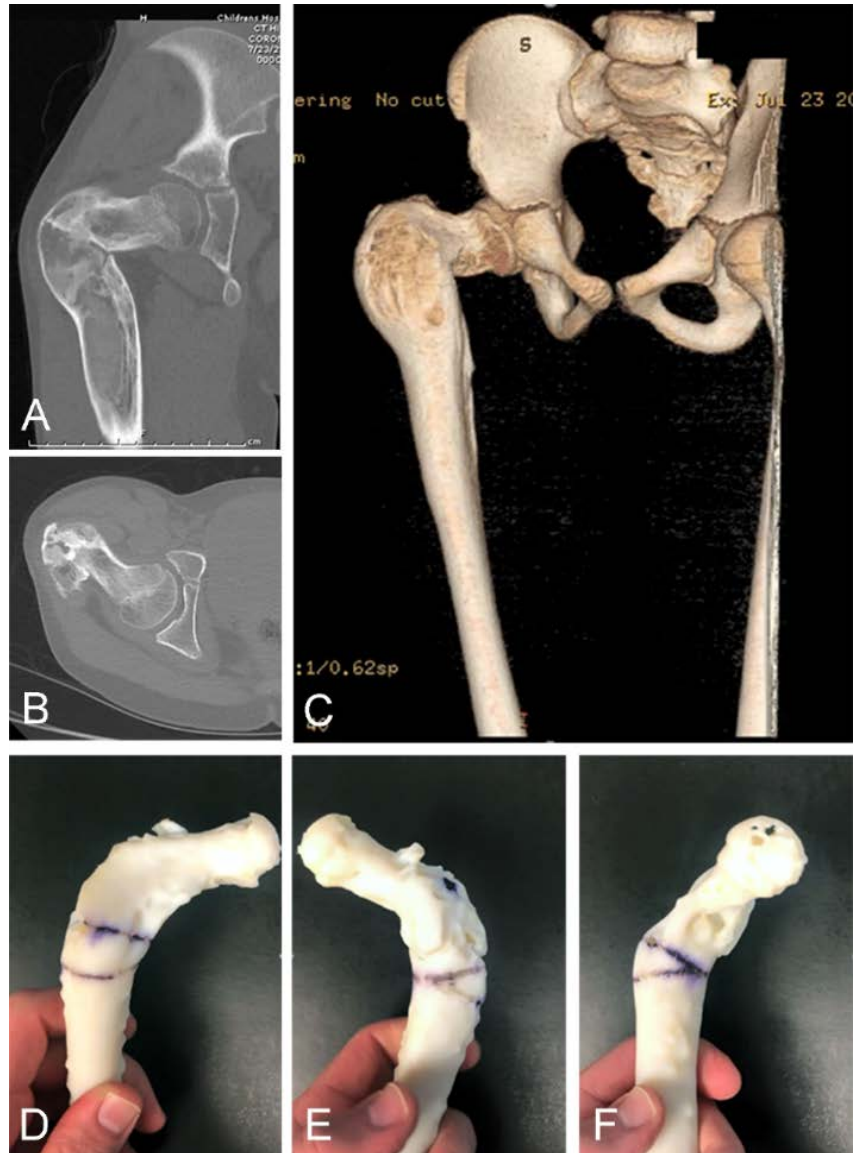


Figure 3. Coronal (A), axial (B), and 3D reconstruction CT scans are helpful in preoperative planning. However, 3D-printed bone models are invaluable in determining the osteotomy sites and the amount of correction (D, E, F). See Supplemental Video 1.

Correction of the Proximal Femoral Deformity

Deformities of the proximal femur are one of the hallmark features of fibrous dysplasia. The extent of disease, age of the patient, and high mechanical loads acting on that region, are the main drivers towards the so-called *Shepherd's crook* deformity.⁴⁹

Ippolito et al. have classified proximal femoral fibrous dysplasia in six types, aiming to predict progression and guide treatment.⁵⁰

- **Type 1** (24%): FD limited mostly to the neck and trochanteric region; normal neck-shaft angle (120-140°)
- **Type 2** (6%): FD limited mostly to the neck and trochanteric region; proximal femoral valgus deformity (>140°)
- **Type 3** (7%): FD limited mostly to the neck and trochanteric region; coxa vara with neck-shaft angle <120°
- **Type 4** (20%): Lateral bowing of the proximal femoral shaft; normal neck-shaft angle (120-140°)
- **Type 5** (14%): Lateral bowing of the proximal femoral shaft; proximal femoral valgus deformity (>140°)
- **Type 6** (29%): Lateral bowing of the proximal femoral shaft; coxa vara with neck-shaft angle <120°

As the classification distribution highlights, the deformity of the femur will not be limited to the proximal aspect. Furthermore, when planning to correct the proximal femur deformity, one needs to take into consideration the presence of other associated deformities. In their series, Types 1 and 2 developed pain but no progressive deformity and were successfully managed with proximal femur fixation alone. On the other hand, Types 3 through 6 had a high rate of progression and recurrence, and needed more extensive surgeries, with “total bone fixation.” Type 6 is the most extensive disease and represents a true *Shepherd’s crook* deformity.

Author’s Preferred Approach (*Shepherd’s crook* deformity)

The goal of surgical correction for *Shepherd’s crook* deformity is to (1) re-establish normal biomechanical forces around the hip and proximal femur, (2) prevent deformity progression, and (3) improve pain and gait

pattern. The main indications for the procedure are pain (continuous, at rest or with walking), and progressive deformity (evident by serial radiographs). If possible, it is preferable to delay surgery until closer to skeletal maturity to minimize the need for revisions due to skeletal growth.

The goals of the surgery are:

1. Achieve neck-shaft angle >120°
2. Correct the lateral bowing of the femur (if present)
3. Correct any sagittal plane femoral deformity (usually anterior apex at midshaft)
4. Restore the rotational alignment and version (typically retroversion)
5. “Total bone fixation” with an intramedullary, load-sharing device along the entire femoral length, with fixation points in the femoral neck and engaging into the epiphysis (Types 3 through 6)
6. Address cystic degeneration of FD (if present), particularly in the femoral neck and intertrochanteric region. Synthetic calcium phosphate bone substitute is the authors’ preferred graft in these instances.

Accurate preoperative planning is crucial for success, as this is a complex multi-directional deformity. Failure to correct all deformities might lead to a reverse or secondary deformity and increase the risk of complications. It is important to obtain full-length films of bilateral lower extremities and 3D imaging of the proximal femur with either CT or MRI. 3D reconstruction images are extremely helpful, and if there is availability, 3D printed models can be a great tool for preoperative planning, and intraoperatively guidance (Figure 3, Supplemental Video 1).

A novel approach for preoperative planning in difficult cases is to simulate the deformity on computer-generated models and determine the exact level of osteotomies and the amount of correction. Based on these simulations, 3D-printed osteotomy templates are made to intraoperatively guide the osteotomy path and

correction, which not only reduces the operative time, but also fine-tunes the correction to achieve the optimal alignment.⁵¹

The expected intraoperative challenges include poor bone quality and lack of normal anatomy, including the absent femoral medullary canal. Intraoperative reduction maneuvers to achieve adequate alignment can lead to intraoperative pathologic fractures and loss of reduction.

One “tip of the trade” is to avoid forceful bone manipulation, especially utilizing sharp bone clamps. The authors recommend that the proximal femur osteotomy be performed after the starting point for the intramedullary implant is secured (lateral entry

nails are preferred). Then, the osteotomy should be provisionally fixed with a 1/3 tubular locking unicortical plate to minimize the need for tight bone clamps (Figures 2 and 4). Bone clamps can still be used over the plate but will need less force to hold the reduction while the canal is being prepared/reamed for implant fixation. The plates are usually removed after the implant is in place. Since FD usually returns after excision, the bigger the nail, the better it is to achieve improved mechanical force and canal filling.

Ideally, the correction is achieved with a single proximal level osteotomy via wedge resection. Adding another proximal level osteotomy makes it very challenging to secure the reduction. If there is a distal femoral deformity, however, another osteotomy will be needed to pass the fairly straight nail through the curved bone

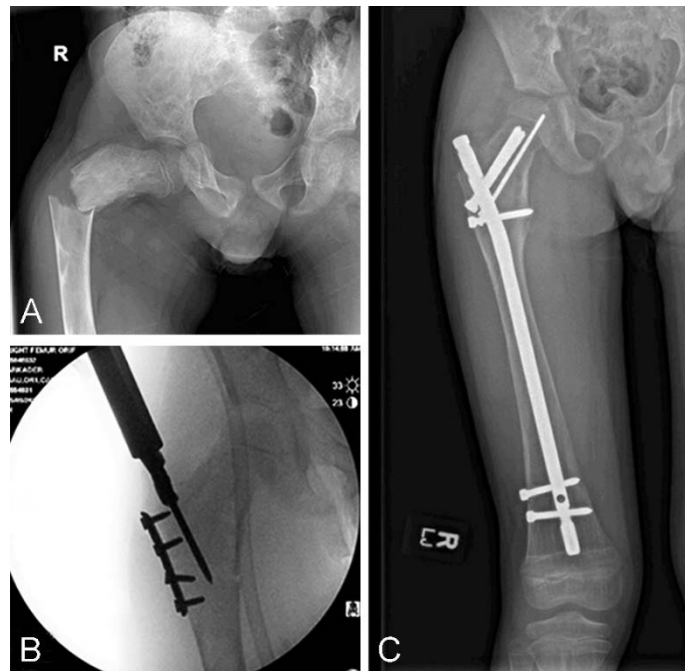


Figure 4. A 7-year-old boy with polyostotic fibrous dysplasia and a femoral fracture (A). A locking plate with unicortical screws was used to provisionally hold the reduction while the femur is being prepared and reamed (B), which was removed after the nail was inserted. Note that supplemental femoral neck fixation was performed with free-hand technique placement of cannulated screws. The patient is pain-free at 1-year follow-up (C).

(usually sagittal plane). These distal level osteotomies can be safely done percutaneously without a wedge resection.

Fibrous dysplasia tends to spare the epiphysis. Therefore, proximal femoral fixation should cross the physis, especially if the remaining growth is not significant. If size permits, fixation devices with two reconstruction screws options are preferred (Figure 1, 2, 4). If there is not enough “real estate” for 2 points of fixation through the neck, a free-handed cannulated screw along the neck, parallel to the reconstruction screw, can augment femoral neck stability (Figures 1, 4).

Quality of Life

While FD has been studied extensively from the medical standpoint, the quality of life of the affected patients and their social and emotional function have only recently come into the spotlight. Kelly et al. reported pain as a common feature in FD, which, regardless of the disease severity, is more prevalent and severe in adults.⁴ Also, inadequate pain control is a factor negatively affecting the patients’ health-related quality of life. The degree of physical impairment of patients with FD is correlated with disease severity and is also comparable to that of patients with similar chronic diseases. Interestingly, patients with FD seem to be “well-adjusted” to their condition and demonstrate mental health scores equal to those of the general U.S. population.⁵² Majoor et al. explored illness perception in 97 patients with fibrous

dysplasia.⁵³ They observed that not only illness perception is affected in FD, but these perceptions are also associated with impairments in quality of life and suggested identifying and addressing maladaptive illness perception in order to improve quality of life.

Summary

Fibrous dysplasia is a benign, non-inherited condition that can present as monostotic, polyostotic, or associated with multisystem disorders. Mutations of the GNAS gene are responsible for the development of FD. Pain, deformity, and pathologic fractures are the most common reasons for orthopaedic intervention. Medical treatment, including pain management, and bisphosphonate therapy, has shown to be effective in some patients. Surgical treatment is indicated when pain is not responsive to medical treatment, a severe or progressive deformity is present, a pathologic fracture has happened or is impending, or there is a high suspicion for malignant transformation. Surgical treatment with intramedullary, load-sharing devices is the recommended approach, aiming at a “total bone fixation.” In complex deformities, novel technologies, including 3D printing and patient-specific guides or hardware should be part of the armamentarium.

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