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CASE REPORT

Fibrous Dysplasia with Secondary Aneurysmal bone Cyst in Long Bones of a Young Girl

Rishabh Bhattacharya[1], Kamal Preet Palta[2], Bishash Roy[3]

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ABSTRACT

Introduction: Fibrous dysplasia (FD) is a bone disorder in which fibrous tissues replace the natural bone matrix. FD can affect any bone in the body; however, it most commonly affects flat and long bones. Aneurysmal bone cysts (ABCs), often described as "blood-filled cysts," are a benign (non-cancerous) type of bone disease. The tibia and femur are among the bones most commonly affected by ABCs. Concurrent FD with secondary ABC in long bones is very rare.

Case Report: We present a case of a 9-year-old girl, who presented to the emergency with pain in her left groin and an inability to bear weight on her left leg. Complete case history was recorded and a preliminary medical examination was performed. Based on radiological findings and non-contrast computed tomography scans, the subject was diagnosed with FD and a concurrent bone cyst. The MRI showed a large fluid–fluid level in the proximal part of the lesion, indicating FD with ABC. Surgical curettage and debridement, followed by plating and bone grafting were planned. Post-operation biopsy performed on the curetted materials confirmed concurrent FD with ABC.

Discussion: Concurrent FD with ABC is rare. After through literature review, we could identify a few reported cases of concurrent FD with ABC in long bones.

Conclusion: Concurrent FD with ABC-like changes is best visualized on MRI, which can reveal conventional fluid–fluid levels, aiding in accurate diagnosis and treatment planning.

KEYWORDS

• Aneurysmal bone cyst • Bone cysts • Femur • Fibrous dysplasia • Long bones

AUTHOR'S AFFILIATION:

[1] Post Graduation 2nd Year Resident, Department of Emergency Medicine, Max Smart Super Speciality Hospital, Saket, Delhi 110017, India.

[2] Principal Consultant and Head of Department, Department of Emergency Medicine, Max Smart Super Speciality Hospital, Saket, Delhi 110017, India.

[3] Senior Consultant, Department of Emergency Medicine, Max Smart Super Speciality Hospital, Saket, Delhi 110017, India.

CORRESPONDING AUTHOR:

Kamal Preet Palta, Principal Consultant and Head of Department, Department of Emergency Medicine, Max Smart Super Speciality Hospital, Saket, Delhi 110017, India.

E-mail: Kamal.Palta@maxhealthcare.com

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INTRODUCTION

Fibrous dysplasia (FD) is a rare and non-hereditary congenital disorder in which normal osteocytes in a healthy bone are replaced by an abnormal fibrous (scar like) tissue. In FD, the fibrous tissue significantly weakens the affected bone over time, making it prone to pathological fractures and causing deformities in the shape of the bone. FD is usually diagnosed in children and young adults.[1] Typically, FD is caused by mutation of a gene, namely GNAS, in the fetus during its development in the womb.[2] Changes in the normal gene prevent bone forming cells from maturing, producing abnormal fibrous tissues in certain bones. Since the gene change occurs while a fetus is developing, only specific bones can be affected by FD. Notably, FD does not spread from one bone to another. FD can affect any bone in the body; however, for some people, FD occurs in the bones on one side of the body.[1,3] The most common bones affected by FD are as follows: Skull and facial bones; Femur, tibia, and fibula; Humerus; Pelvis; Ribs.[3]

FD occurs in two forms:[2]

1. Monostotic, which affects one bone and is active while the child is growing but often becomes inactive after puberty.
2. Polyostotic, which affects multiple bones and may remain active throughout a person's life.

Polyostotic dysplasia can further be divided into the following types:

- McCune-Albright syndrome affects the bone, skin (café-au-lait lesions), and endocrine tissues.[4]
- Mazabraud syndrome affects soft tissues, causing intramuscular myxomas.[5]
- Jaffe-Lichenstein syndrome also forms café-au-lait lesions; however, it can be distinguished from McCune Albright syndrome by lack of endocrinopathies.[6]
- Osteofibrous dysplasia is a rare form of FD that primarily affects the tibia and is confined to the cortices.[7]
- Non-orthopedic manifestations involve severe craniofacial deformities leading to cosmetic deformities, blindness, hearing loss, and airway obstruction.[8]

The incidence of FD is 1 in 5,000 to 10,000.[9] FD often causes discomfort but can sometimes show as a painless lump in the maxillofacial or cranial regions. Most cases of FD are self-limiting, showing stabilization after puberty. Pain is the most prevalent manifestation of FD in adult patients compared to the paediatric population, with discomfort at the involved location occurring in most cases. The majority of subjects suffering from FD, particularly those with monostotic type dysplasia, show no symptoms.

Aneurysmal bone cysts (ABCs) are benign and expansile lytic bone lesions with multiple thin-walled interconnected cystic cavities that contain blood.[10] ABCs can cause aggressive bone disintegration, accounting for approximately 2% of bone tumours.[11]ABC occurs due to deregulation of the USP6 gene, affecting the bone characterized by classical imaging findings and is neither an aneurysm nor a cyst.[12] ABC-like changes, or secondary ABCs, are not caused by uncontrolled cell growth or tumor development that is often associated with conditions, such as chondroblastoma, osteoblastoma, non-ossifying fibroma, and giant cell tumors. Note that ABC-like changes are not linked to deregulation of the USP6 gene.[13]

CASE REPORT

A 9-year-old female presented to the emergency with pain in her left groin and an inability to bear weight on her left leg for the past two days. Two days ago, the subject missed a step and tripped while descending from stairs. The subject also complained about experiencing intermittent pain in her left groin after a history of a fall while riding a bicycle nine months ago. After that fall, the subject gradually developed a limp and has been on and off analgesics ever since. On physical examination, the subject was conscious, alert, and cooperative. The left leg was in an externally rotated position. The subject had tenderness over the left groin on palpation, with decreased range of motion. Her vitals were within normal limits. The rest of the general head-to-toe examination revealed no significant abnormal findings. An X-ray of the left femur was done for the patient. A ground glass appearance with a fracture in the proximal femur can be observed in the X-ray radiograph (*Figure* 1).

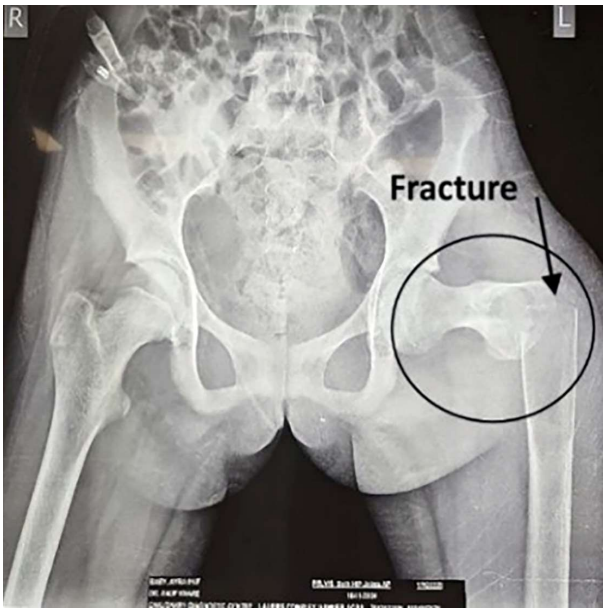

Figure 1: Radiograph obtained during initial preliminary examination of the left femur of the subject

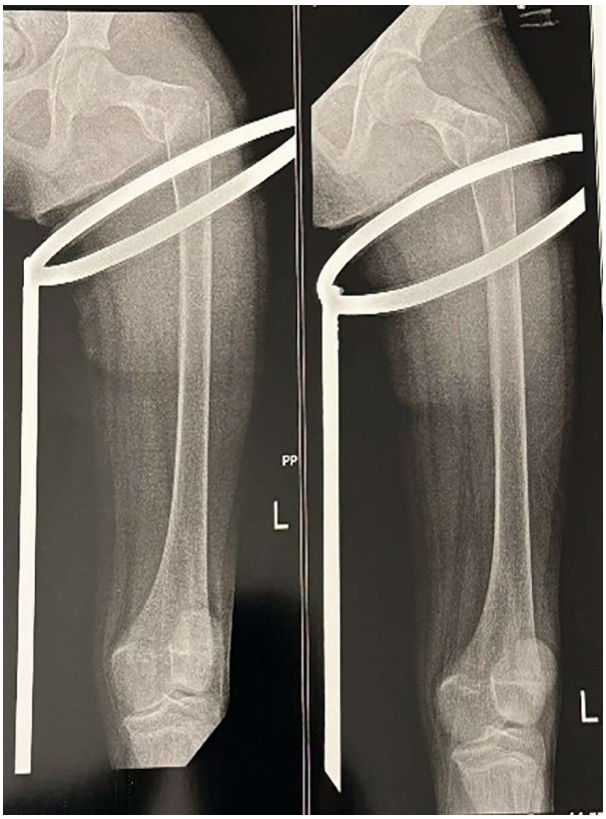

Figure 2: Radiograph obtained after thomas splint was applied to the left femur of the subject

A Thomas splint was applied (*Figure* 2), and non-contrast computed tomography (NCCT) scans of the pelvis and femur were done. In the NCCT scans (Figure 3), a pathological fracture in the proximal diaphysis of the left femur and a well-defined expansile central osteolytic lesion measuring 3.3 × 2.6 × 17.3 cm^3 were observed in the proximal 1/3rd of the left femur, with a thinned-out cortex and isodense soft tissue, indicating fluid content in the cystic lesion. The NCCT scans indicated the presence of a solitary bone cyst with a pathological fracture.

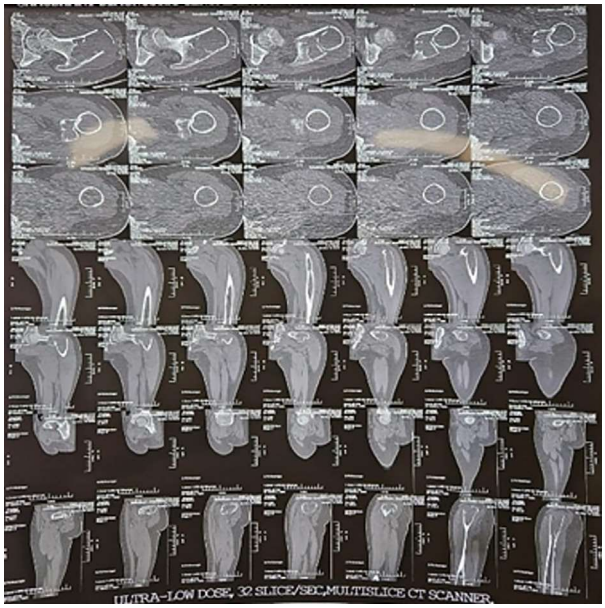

Fig 3. NCCT scans recorded from the left femur of the subject

Based on the initial survey scans and physical examination, the subject was diagnosed with FD and was referred for surgical management. The subject was planned for curettage of the left proximal femur and open reduction and internal fixation (ORIF) with plating and bone grafting. Additionally, biopsy was planned on the curetted material.

Before the surgery, magnetic resonance imaging (MRI) of the left femur was also done. The MRI (*Figure* 4) showed a well-defined expansile lytic lesion in the proximal meta-diaphyseal region of the left femur and a pathological fracture through the lesion in the intertrochanteric region. The MRI also showed a large fluid–fluid level in the proximal part of the lesion. The MRI findings indicated the presence of a simple bone cyst with a pathological fracture, along with adjacent hematoma and muscular and intermuscular edema. Based on the MRI findings, a final diagnosis of monostotic FD with ABC-like changes and a pathological fracture was made.

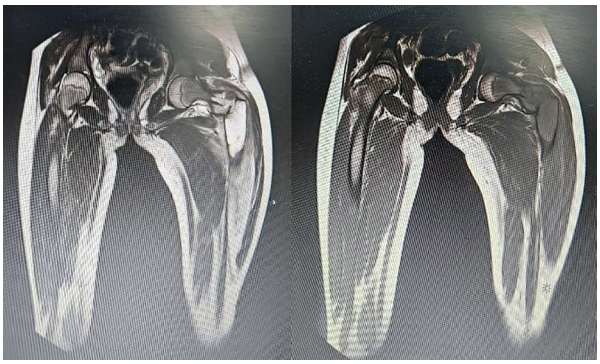

Figure 4: MRI images showing the right and left femurs of the subject

After clearance from the anaesthesiologist, the patient was moved to the operation theatre for the planned surgery. Under general anaesthesia, bone curettage and debridement were performed. After curettage, the bone cavity was filled with a synthetic bone graft (fraclink and gene-x), and the fracture was reduced. Then, a PHILOS plate having 8 holes was inserted and fixed with the help of screws (Figure 5).

Finally, biopsy was performed on the curetted material. The biopsy showed sections having variably mineralized bony trabeculae consisting of mature cortical bones. Some sections also showed enchondral ossification. Inter trabecular spaces were predominantly haemorrhagic. Additionally, few haemorrhagic cystic fragments palisaded by many osteoclast-like giant cells and lymphomononuclear cells were observed. The biopsy findings indicated the presence of a bone cyst with ABC-like areas. Furthermore, the biopsy confirmed the absence of malignancy. The subject was confirmed having FD with ABC.

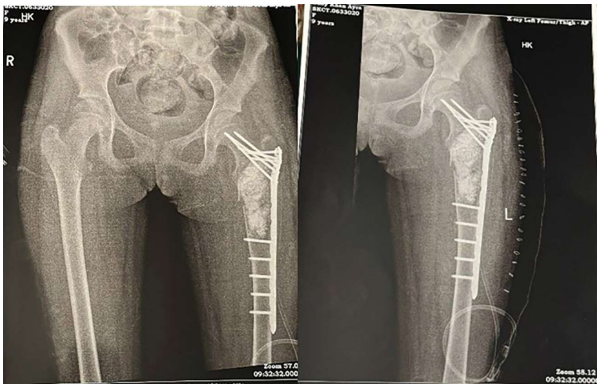

Figure 5: Radiograph obtained after completion of the surgery on the left femur of the subject

DISCUSSION

Concurrent ABC-like changes are uncommon and are often observed in craniofacial FD. Only a few examples have been described when FD in the appendicular skeleton coincided with ABC-like changes.[14] Concurrent occurrence of FD and ABC is extremely rare, with earliest report presented by Buraczewski *et al*. in 1971.[15] Typically, hemangioma, osteoblastoma, osteoclastoma, and osteosarcoma are the main conditions that are associated with ABC. On literature review, we came across some cases of concurrent FD with ABC-like changes that affected various bones in the body, primarily the skull bones.[15,16,17] However, only a few cases of FD with ABD in long bones have been previously reported, with patients presenting a wide range of signs and symptoms, the majority of which were complaints of pain in the affected body part.[10] Radiograph or CT scan evaluation can miss the concomitant ABC-like changes that are best depicted on MRI with classical fluid–fluid levels.[18]

CONCLUSION

Concurrent FD with ABC-like changes is rare, with most reported cases involving the craniofacial bones. On radiographs or CT scans, concurrent FD with ABC-like changes may not be detected or may be misdiagnosed. Concurrent FD with ABC-like changes is best visualized on MRI, which can reveal conventional fluid–fluid levels, aiding in accurate diagnosis and treatment planning. A better understanding of concurrent occurrence of FD with ABC-like changes can reduce the risk of any unintended problems during surgery.

Conflict of Interest: There are no conflicts to declare.

REFERENCES

1. Tafti D., Cecava N.D. Fibrous Dysplasia. In: StatPearls. StatPearls Publishing, Treasure Island (FL) 2023; PMID: 30422542.
2. Arkell C., van Heerden J. Fibrous dysplasia: a current concepts review. SA Orthopaedic Journal 2023; 22(4): 208-17.
3. Singh G., Walizai T., Bell D., *et al*. Fibrous dysplasia. Reference article, Radiopaedia.org 2025; rID: 4915.

4. Galván M., Herdenes D., Pérez M., Pacheco D., Suárez J., Cantillo P., Pérez D.Z., Caycedo. Comprehensive Multidisciplinary Insights into Fibrous Dysplasia and McCune-Albright Syndrome, International Journal of Science and Research 2025, 14(1): 323-29.

5. Gaillard F., Campos A., Bell D., *et al*. Mazabraud syndrome. Reference article, Radiopaedia.org 2025; rID: 6569.

6. Henriques J.C., Mitri F.F., De Paulo L.F., Barbosa G.L., Batista J.D., Cardoso S.V., Loyola A.M. Jaffe-Lichtenstein Syndrome Associated with Simple Bone Cyst. Oral Surgery, Oral Medicine, Oral Pathology and Oral Radiology. 2020 Jan 1; 129(1): e26.

7. Liu R., Tong L., Wu H., Guo Q., Xu L., Sun Z., Yan H. Osteofibrous dysplasia: a narrative review. Journal of Orthopaedic Surgery and Research. 2024 Mar 27; 19(1): 204.

8. Menon S., Venkatswamy S., Ramu V., Banu K., Ehtaih S., Kashyap V.M. Craniofacial fibrous dysplasia: Surgery and literature review. Annals of maxillofacial surgery. 2013 Jan 1; 3(1): 66-71.

9. Ha-Young K., Shim J.H., Heo C.Y. A Rare Skeletal Disorder, Fibrous Dysplasia: A Review of Its Pathogenesis and Therapeutic Prospects. International Journal of Molecular Sciences. 2023 Nov 1; 24(21): 15591.

10. Wahab H., Birlas A., Ibrahim M., Khattak N. S, Qadir R.I. Case report of a Young Girl with Fibrous Dysplasia (FD) Concomitant with Secondary Aneurysmal Bone Cyst (ABC).J Pak Orthop. Assoc. (JPOA) 2023; 35(2).

11. Shamala S., Beeula A., Devi M., Hariganesh P. A large aneurysmal bone cyst of mandible: A rare case report. Journal of Oral and Maxillofacial Pathology. 2023 Jul 1; 27(3): 548-52.

12. Oliveira A.M., Hsi B.L., Weremowicz S., Rosenberg A.E., Cin P.D., Joseph N., Bridge J.A., Perez-Atayde A.R., Fletcher J.A. USP6 (Tre2) fusion oncogenes in aneurysmal bone cyst. Cancer research. 2004 Mar 15; 64(6): 1920-3.

13. Ricardo R., Zahrah D., Pelaez L., Temple H.T., Murakami J.W. Update on aneurysmal bone cyst: pathophysiology, histology, imaging and treatment. PediatrRadiol. 2022; 52: 1601-14.

14. Alawi F. Benign fibro-osseous diseases of the maxillofacial bones: a review and differential diagnosis. Pathology Patterns Reviews. 2002 Dec 1; 118 (suppl_1): S50-70.

15. Buraczewski J., Dabska M. Pathogenesis of aneurysmal bone cyst: relationship between the aneurysmal bone cyst and fibrous dysplasia of bone. Cancer. 1971 Sep; 28(3): 597-604.

16. Lee J.W., Kim J.H, Han S.H., Kang H.I. Fibrous dysplasia with aneurysmal bone cyst presenting as painful solitary skull lesion. Journal of Korean Neurosurgical Society. 2010 Dec; 48(6): 551.

17. Diercks R.L., Sauter A.J., Mallens W.M. Aneurysmal bone cyst in association with fibrous dysplasia. A case report. The Journal of Bone and Joint Surgery. British volume. 1986 Jan; 68(1): 144-6.

18. Spinnato P., Colangeli M., Pedrini E., Parmeggiani A., Papalexis N., Crombé A., Gambarotti M., Bazzocchi A. Aneurysmal bone cyst-like changes developed in melorheostosis with epiphyseal osteopoikilosis. Skeletal Radiology. 2024 Jul; 53(7): 1437-41.