CASE REPORT

# Clinicoradiologic perspective of a severe case of polyostotic fibrous dysplasia

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## ABSTRACT

Fibrous dysplasia (FD) of bone is a congenital non-heritable disorder that was first reliably reported by von Recklinghausen, when he described patients with pathologic condition of bone characterized by deformity and fibrotic changes that he termed as osteitis fibrosa generalisata. FD may involve one bone (monostotic) or multiple bones (polyostotic) and occurs throughout the skeleton with predilection for long bones, ribs, and cranio-facial bones. Seventy percent of the lesions are monostotic and asymptomatic, and identified incidentally. The polyostotic form of disease is often deforming and devastating, with multiple skeletal complications like repeated fractures, limb length discrepancies, and bone pains. The bone lesion of unknown origin is characterized by slow progressive replacement of normal bone by abnormal proliferative, isomorphic fibrous tissue. This case report documents a 40-year-old male with severe polyostotic FD that involved most of the skeleton, including long bones of all extremities, pelvis, facial bones, and skull base. Initial evaluation consisted of physical examination, plain radiographs, which was followed by computed tomography scan, Single-photon emission computed tomography scan, and biochemical and hematological examination. This paper stresses on the clinical implications and management of this rare debilitating disease.

***Key words:*** 3D volume rendered image, ground glass appearance, polyostotic fibrous dysplasia, serum alkaline phosphatase, Shepherd Crook deformity

## INTRODUCTION

Fibrous dysplasia (FD) is a slowly progressive condition characterized by replacement of normal bone with an amalgamate of cellular fibrous tissue and irregular bony trabeculae.[1] First medical documentation made by von Recklinghausen in 1891[2] was followed by recognition of the lesion as a separate entity by McCune and Bruch in 1937, immediately succeeded by coining of the term "fibrous dysplasia" by Lichenstein in the following year.[3]

Categorized according to the number of involved bones into monostotic and polyostotic variants and as McCune-Albright and Jaffe-Lichenstein syndrome owing to endocrinal association, the lesion exhibits a wide range of clinical and investigative features.[1] Polyostotic FD has been found to localize in most cases to long bones, ribs, and skull, occurring unilaterally in most cases[2] with involvement of skull bones such as ethmoid, sphenoid, frontal, maxillary, temporal, and occipital bones in decreasing order of frequency.[4] Computed tomography is the favored imaging modality for the lesion, as it provides a far superior insight into the details of the texture and extent of the lesion.[4] With improved understanding of the molecular level details of mutations of $G_s\alpha$ at R201 and more recently at Q227,[5] the perplexing lesion may soon be unraveled.

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| Quick Response Code: | Website: www.jomfp.in |
| | DOI: 10.4103/0973-029X.99097 |

## CASE REPORT

A 48-year-old male patient was seen in consultation for a lesion in the proximal femur. He complained of chronic, mild-to-moderate pain in his left hip and walked with a noticeable limp. Physical examination revealed hip deformity and minimal limb length discrepancy. Further clinical examination revealed frontal bossing, distortion of skull shape, and facial asymmetry [Figure 1]. No cutaneous lesions were observed. The patient had twitching on left side of the face.

Conventional radiography was performed in relation to skull, long bones of the lower extremities, chest, and hand. The radiographs of skull showed diffuse sclerosis with widening of the skull table in right frontal region, resulting in frontal bossing. It also showed obliteration of paranasal sinuses and an exophytic lesion of the right mandible. Radiographic evaluation of the region of left hip joint, femur, and pelvis revealed multiloculated lytic lesions in the iliac ramus blade and femur, which was partially surrounded by a sclerotic rim. Shepherd Crook deformity was seen. Radiograph of chest showed multiple expansile lytic lesions involving both anterior and posterior ends of ribs of both sides [Figure 2].

Further, detailed radiological investigations were performed using computed tomography (CT), and 3D volume rendered images of the craniomaxillofacial region were obtained. Plain CT, axial section (bone window), showed thickening of frontal, parietal, occipital, and basosphenoid regions and enlargement of occipital bone, right mandibular ramus, and left mastoid bone [Figure 3]. 3D volume rendered images of the craniomaxillofacial region showed deformation of the left and right hemicranium, affecting fronto-temporo-parietal bones along with bony expansion of the external occipital protuberance. Floor of anterior and middle cranial fossa was involved. Thickening of the roof of orbit, frontal bones, and frontonasal process was seen along with obliteration of maxillary sinus, and an expansile bony lesion was noted on the right mandible [Figure 4]. Biochemical investigations were performed to rule out associated endocrinal abnormalities [Table 1]. Serum levels for alkaline phosphatase, calcium, and PO4 were normal. Detailed endocrinological tests, including thyroid and parathyroid function tests, and serum hormone levels (ACTH, growth hormone) were performed and normal values were obtained.

Due to a previous history of an allergic reaction to medically used radioisotopes, technetium scintigraphy was not performed. Although biopsy was contemplated, the patient refused any further investigative procedures and treatment.

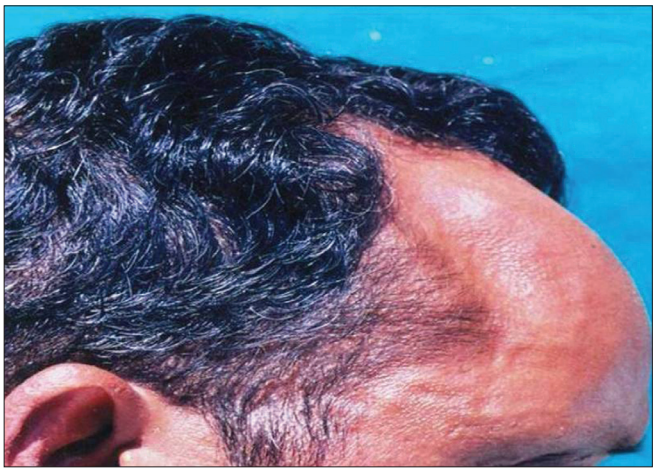

**Figure 1:** Cropped profile picture of the patient showing frontal bossing

Differentially diagnosed from ossifying fibroma, Paget's disease of bone, aneurysmal bone cyst, and giant cell tumor (brown tumor of hyperparathyroidism), the patient was treated with Alendronate 70 mg once a week for 6 months and was evaluated thereafter to find a marked diminution in the chief presenting complaint and associated pain. The patient is under observation for the last 4 years now and there is a marked improvement in associated symptoms.

## DISCUSSION

FD is an idiopathic skeletal disorder in which medullary bone is replaced by poorly organized, structurally unsound fibro-osseous tissue.[1] Most patients are under 30 years of age at the time of diagnosis. The majority of cases (75% to 80%) are limited to the monostotic fibrous dysplasia (MFD), which most often involves the ribs or femur. The craniofacial bones may be involved in MFD in up to 25% of cases; most commonly affected are the maxilla and mandible. Polyostotic FD (PFD) comprises 20% to 25% of all cases. Usually, there is unilateral bone involvement, but in severe cases bilateral disease can occur. The craniofacial bones are more often (40% to 50%) involved in PFD.[2] Albright's syndrome consists of PFD plus pigmented skin macules and sexual precocity. This syndrome is relatively rare. It is 40 times less common than MFD, and it occurs almost exclusively in females.[5] Patients with craniofacial FD involvement usually present with asymmetric cheek swelling. Encroachment into the paranasal sinuses, nasal fossae, orbit, or neurovascular canals can lead to nasal obstruction, headaches, and visual disturbances.[6]

Extensive maxillary involvement results in facial distortion, referred to as leontiasis ossea ("lion face"). New FD lesions usually do not develop after the growth plates have fused. Some lesions, however, continue to grow after skeletal maturation. FD is usually a benign condition, which begins in childhood and stabilizes at puberty. About a third of patients

**Table 1: Blood biochemical investigations**

| Test | Value obtained | Normal value |
|---|---|---|
| Serum $T_3$ | 1.2 ng/mL | 0.8–2.0 ng/mL |
| Serum $T_4$ | 8.0 µg/mL | 4.66–13.2 µg/mL |
| Serum TSH (RIA method) | 2.8 µIU/mL | 0.27–3.75 µIU/mL |
| Parathyroid hormone | 25.0 pg/mL | 12.0–72.0 pg/ml |
| Leutinizing hormone | 7.2 mIU/mL | 1–15 mIU/mL |
| Follicular stimulating hormone | 82 mIU/mL | 1–15 mIU/mL |
| Serum creatinine | 1.2 mg/dL | 0.7–1.5 mg/dL |
| Serum alkaline phosphatase | 427.0 µ/L | 60–170 µ/L (Adult) |
| Serum calcium (total) | 9.2 mg/dL | 8.5–11.0 mg/dL |
| Serum phosphorus | 4.5 mg/dL | 2.5–5.0 mg/dL (Adult) |
| Urine hydroxyproline levels | Not Evaluated | - |

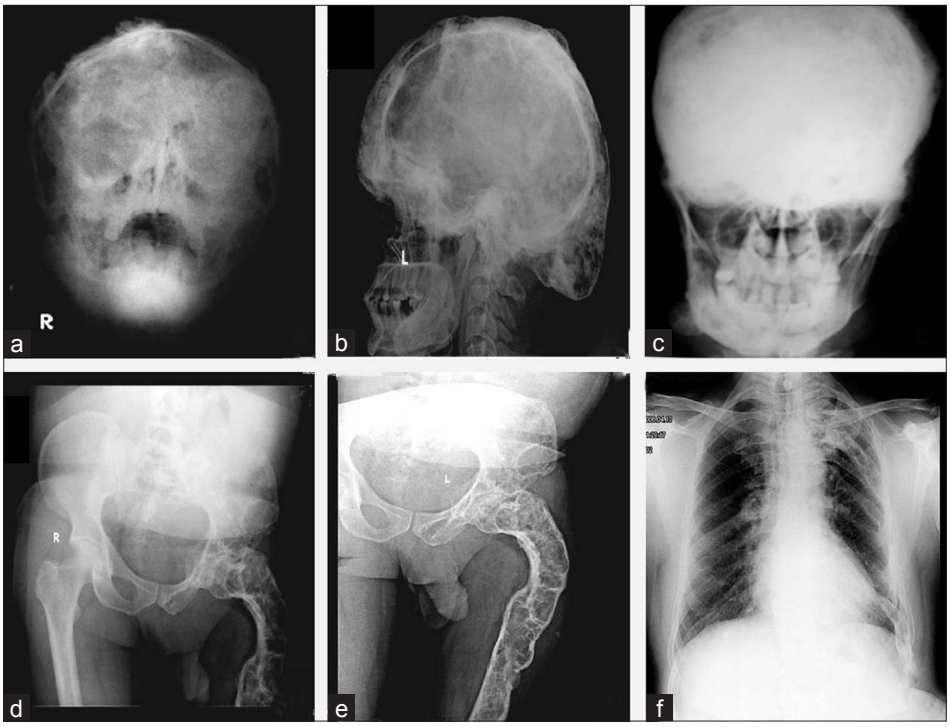


**Figure 2:** Conventional radiography (a) X-ray PNS face showing widening of the skull table in the right frontal region and obliteration of paranasal sinuses with areas of diffuse ground glass haze. (b) Lateral view skull showing diffuse sclerosis with thickening of skull vault and frontal bossing. Diffuse lucent areas are also seen and frontal and maxillary sinuses are obliterated. (c) PA view skull showing obliteration of frontal and ethmoidal sinuses with areas of diffuse ground glass haze. Exophytic lesion on the right mandible. (d) Left hip joint and pelvis showing multiloculated lytic lesions in the iliac ramus blade. (e)X-ray left femur showing a large elongated Intramedullary multiloculated expansile lesion reaching the head of left femur. Typical coxa vara abnormality – Shepherd Crook deformity (lateral bowing) is seen. Lesion is partially surrounded by a sclerotic rim. (f) X-ray chest (PA view) showing multiple expansile lytic lesions involving both anterior and posterior ends of the ribs of both the sides

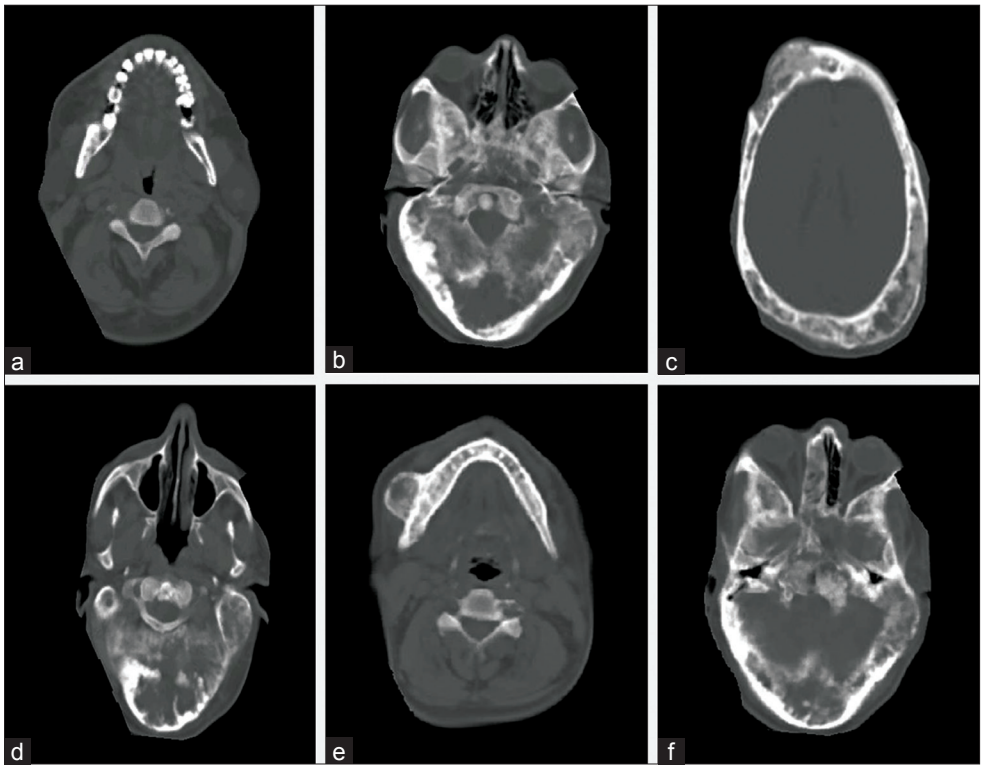


**Figure 3:** Conventional tomography: Plain CT axial section (bone window) showing (a) enlarged madibular ramus. (b) occipital bone and basosphenoid with cotton wool appearance. (c) thickened frontal and parietal bones with amorphous density. (d) enlarged occipital bone and left mastoid.(e) exophytic lesion of the right madibular ramus. (f) obliteration of the right half of ethmoid sinus

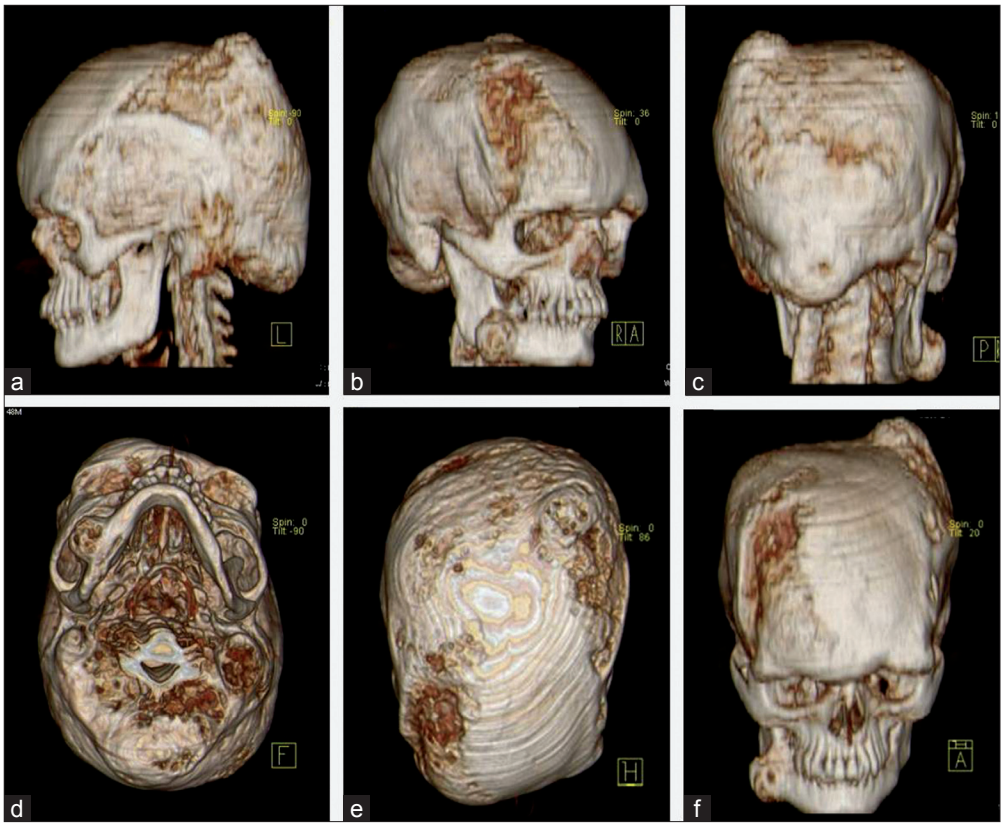


**Figure 4:** 3D Volume Rendered Images of the skull and face showing bony deformity of (a) left hemicranium involving frontotemporo-parietal bone and the floor of anterior and middle cranial fossa. Zygomatic bone and left mandible are normal. (b) right hemicranium and changes on the right side of vault. There is thickening of frontal bones, frontonasal process and obliteration of maxillary sinus. Roof of orbit is thickened. Expansile bony lesion on the right mandible. (c) Bony lesion causing overgrowth of external occipital protuberance. (d) Expansile lesion involving the mandible and left mastoid. (e) Frontal and Parietal thickness, cortical erosions in the vault. (f) Expansion of mandible and parietal bones

have further progression in adulthood.[7]

The European Pediatric Orthopedic Society has performed a multicenter clinico-pathologic study to gain insight into the natural history of FD. Fifty-three patients from 11 centers were included. Twenty-three patients with a mean age of 15 years had monostotic involvement. Ten, with a mean age of 11 years, had polyostotic involvement, and 20 with a mean age of 4.5 years had McCune-Albright syndrome. In the cohort with monostotic disease, the most common site of involvement was the femur. Lesions in that group presented as an incidental finding or with pain, swelling, or fracture. The other common areas of involvement were the tibia, humerus, ribs, clavicle, and craniofacial skeleton. A majority of the monostotic cases did not progress, and the long-term outcome was usually satisfactory in those cases regardless of treatment.[8]

The radiographic characteristics of FD, as described by Fries in 1957, are pagetoid (56%), a mixture of dense and radiolucent areas of fibrosis; sclerotic (23%), mass if homogenously dense; and cystic (21%), aspherical, or ovoid lucency surrounded by a dense boundary.[9] CT is the study of choice for diagnosis and follow-up because of its superior bony detail and accurate assessment of extent of the lesion. Furthermore, CT can often assist with differentiating FD from other osteodystrophies of the skull base, including otosclerosis, osteogenesis imperfecta, Paget disease, and osteopetrosis.[10] In our case, a CT of craniomaxillofacial region was performed and revealed findings consistent with FD of frontal, parietal, occipital, and basosphenoid region; right mandibular ramus, and left mastoid bone. 3D volume rendered images of the craniomaxillofacial region showed deformation of the left and right hemicranium, affecting fronto-temporo-parietal bones along with bony expansion of the external occipital protuberance. Floor of anterior and middle cranial fossa was involved. Thickening of the roof of orbit, frontal bones, and frontonasal process was seen along with obliteration of maxillary sinus, and an expansile bony lesion was noted on the right mandible.

Although FD is a benign and slowly progressive disorder, in the late stage of the disease, expansion of the cranial bones can cause mass effect on the cranial structures. As in our case, the progressive growth of these lesions may cause difficulties in management.

## CONCLUSION

This is a case of PFD having extensive lesions in

craniomaxillofacial region, hip, femur, and ribs. There were no café-au-lait macules or endocrinal abnormalities. Most lesions of FD are monostotic, asymptomatic and identified incidentally and can be treated with clinical observation. Knowledge of the various appearances, complications, and associations of FD is important to ensure the accurate diagnosis and appropriate management of this disease.

## ACKNOWLEDGMENT

We wish to acknowledge the intellectual support rendered by Dr Hardas Singh Sandhu and Dr. Atul Kapoor, Amritsar, Punjab, India, towards realization of this article in its present form.

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**How to cite this article:** Sandhu SV, Sandhu JS, Sabharwal A. Clinicoradiologic perspective of a severe case of polyostotic fibrous dysplasia. J Oral Maxillofac Pathol 2012;16:301-5.

**Source of Support:** Nil. **Conflict of Interest:** None declared.