

A Case of Craniofacial Polyostotic Fibrous Dysplasia

Justin Clark^{1*}, William Carson²

1. Department of Radiology, Naval Medical Center San Diego, San Diego, USA

2. Department of Radiology, Naval Hospital Okinawa, Chatan, Japan

* **Correspondence:** LT Justin S. Clark, Department of Radiology, Naval Medical Center San Diego, 34520 Bob Wilson Drive, San Diego, CA 92134, USA

(✉) justin.s.clark@med.navy.mil

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ABSTRACT

We present the case of a patient with craniofacial polyostotic fibrous dysplasia. Polyostotic fibrous dysplasia is relatively rare and usually presents in late childhood/early adulthood. It is occasionally associated with endocrine disorders such as McCune-Albright syndrome. The benign pathology of this bone tumor belies its implications in the region of the skull base. Craniofacial polyostotic fibrous dysplasia can have devastating complications depending on which ostia are involved, including vision loss. Our patient was already beginning to experience visual field deficits from ischemic neuropathy. He was treated surgically with optic nerve decompression; however, the efficacy of this approach is currently being debated.

CASE REPORT

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A 32 year old male came to his battalion aid station after he noted swelling to his right cheek, shortly after reporting to his new duty station overseas. This swelling had begun 3 months ago, and had noticeably increased in this relatively short interval. He had no history of trauma or infection to this area. The patient denied headaches, pain to the enlarged area, fevers/night sweats, or congestion/sinus pressure. He did note, however, occasional blurred vision in his right eye in the last few weeks. The patient had not been deployed in the last year, or traveled to any third-world countries.

Upon examination, the patient had a 6x6x2 cm bony prominence to his right zygomatic arch. This prominence was non-tender and non-mobile without overlying skin changes or redness. Initial evaluation of his ears, eyes, mouth, and nose were unremarkable. His voice was of normal quality, and he had full range of motion in his jaw. The patient's neck did not demonstrate thyromegaly or lymphadenopathy and was supple. He did not have any skin lesions. The patient had no other

complaints, had no previous significant medical history, and was otherwise in good health.

His facial bone plain radiograph revealed near complete opacification of right maxillary sinus and a bony prominence in the right frontal/parietal region. (Figure 1) A facial CT was performed several days following for better characterization of the area. This examination demonstrated bony expansion of the skull base, with complete opacification of the right maxillary sinus, extending into the right aspect of the maxilla inferiorly and the right lateral orbital wall, orbital roof and into the right aspect of the frontal bone in the region of the forehead. Associated narrowing of the optic canal was present on the right side which was approximately 3.25mm in diameter. The foramen spinosum and ovale were essentially symmetric with the contralateral side, with no definite evidence of stenosis on this initial examination. The matrix of the lesion was ground glass. Furthermore, disruption of the anterior maxillary wall was noted along with extension into the occipital condyles and a single lesion in his right anterior mandible. (Figures 2,3,4)

The patient was evaluated shortly thereafter by otolaryngology and ophthalmology at our local institution. They agreed that the patient likely had polyostotic fibrous dysplasia, but were struck by the advanced age at onset. The relatively rapid growth of the lesion, 3 months of physical and visual symptoms, and extent of the lesion were also concerning. Based on these factors, they were concerned about malignant transformation and recommended the patient be transferred to another institution for further consultation as soon as possible. An in-depth exam by the ophthalmologist revealed that the patient had abnormal visual field testing with mild general reduction of sensitivity in his right eye, which confirmed the suspicion that he was suffering from ischemic optic neuritis. He was also referred to dentistry for evaluation of the vitality of his right upper molars, as the lesion extending into his maxilla. This evaluation concluded that he was not in danger of any dental complications.

Shortly after admission to the stateside referral hospital, the patient had additional imaging studies to confirm the location and nature of his lesion. A stereotactic CT further characterized his lesion, which was seen to extend from the alveolar ridge/frontal bone to the right anterolateral rim of the foramen magnum with confirmed narrowing of the right optic canal, superior orbital fissure, and foramen ovale. Given the nature of his disease, the referral otolaryngologist recommended urgent biopsy to confirm the diagnosis. A biopsy of his right maxilla showed fibroblast proliferation and hyalinization consistent with fibrous dysplasia, but the sample was sent to another institution for verification. Utilizing the endoscopic technique, his right optic nerve was decompressed and his right maxillary sinus was debulked using the Caldwell-Luc approach. His post-operative course was uncomplicated, and he returned to his overseas duty location. Subsequent testing showed that his visual field general reduction in sensitivity had improved. For medical treatment of his fibrous dysplasia, he was started on alendronate and vitamin D. Our institution's internal medicine department was consulted to see if the patient had an associated endocrine disorder, such as McCune-Albright Syndrome. His thyroid function tests, testosterone, prolactin, insulin-like growth factor, and FSH/LH levels showed no signs of an endocrinopathy. The patient was also referred to dentistry for vitality testing of his right upper molars, as his post-op CT's demonstrated growth of the lesion into his right maxillary alveolus. To complete his work-up for cranial structures that could be affected by his lesion, audiograms did not reveal any associated hearing loss.

Repeat visual field tests have verified that his visual field is stable, and his only complaint is the development of an area of hyperesthesia in the region of his right cheek. A bone scan (Figure 6) showed areas of increased activity in his right humerus and forearm, and plain films of these regions (Figure 7) laid bare additional lesions that have characteristics of fibrous dysplasia. These additional studies confirmed the polyostotic nature of our patient's disease.

DISCUSSION

Fibrous dysplasia of the skull most frequently presents as painless bony enlargement of part of the jaw or face in patients in the first decade of life (1). This case report and several others demonstrate that the age of presentation can be variable, though (2). If the skull base or orbital areas are involved the presentation can be much more concerning with loss of vision or nerve palsies. Pain in the affected area can be associated with fibrous dysplasia lesions in long bones which have subsequent occult fractures (3,4). Skin lesions in the form of café au lait spots point to McCune-Albright syndrome. In this syndrome, these skin findings are associated with endocrine disorders and fibrous dysplasia (1,3,4,5). This benign tumor can masquerade as several other benign lesions, and so the clinical picture is important for differentiation. The mnemonic FEGNOMASHIC can be helpful when considering this diagnosis.

This benign disease of bone accounts for about 2.5 to 10 percent of bone tumors. The disease process is simple and insidious, as normal bone is replaced by fibroblasts. Recent research has linked fibrous dysplasia to a mutation in the Gs gene which is located on chromosome 20q13.2-13.3 (8). The majority of cases are found in the first three decades of life. This disease has an equal distribution between males and females; however, McCune-Albright syndrome has a predilection for females (3).

Plain radiographic images can give some illumination of the underlying process in long bones, pelvis, or ribs but prove inadequate for visualization of the skull. (7) CT is the medium of choice for initial evaluation of suspected bony tumors of the craniofacial area, as it provides better characterization and localization of the lesion (5,6). The often used "ground glass appearance" description for fibrous dysplasia can be misleading. Fibrous dysplasia lesions can just as frequently be seen as purely lytic, sclerotic, mixed/pategoid, or cystic (3,4). MRI, while useful for looking at compression of nearby structures as in this case, is sub-optimal for diagnosis due to the variable pattern of fibrous dysplasia (2,5,6).

Initial evaluation of a suspected case of fibrous dysplasia should include laboratory studies. Elevated serum alkaline phosphatase and urine hydroxyproline are usually noted in the active stage of the disease, to due the extent of bone remodeling (3). These markers can also be used to track treatment progress (3). Checking levels of testosterone, estradiol, and estrone are also useful when ruling out McCune-Albright syndrome (7). Biopsy and histological examination of lesions is often necessary when the diagnosis is unclear or when concerned for malignancy, as in our case. Gross examination of lesions reveals fibrous, tan, and gritty tissue. Hemorrhage or cystic changes are frequently noted within (7). Microscopic characteristics include disorganized spindles of bone, which have been compared to Chinese letters or alphabet soup, and foci of cartilage (3,6). Cells within the lesion will be largely fibroblasts with an immature and poorly differentiated appearance (7).

Surgical treatment is usually necessary for lesions that have begun to encroach upon critical structures. Timing and

necessity of optic nerve decompression continues to be debated in light of an unclear correlation between optic canal stenosis and vision loss (8). A study by Tan revealed that surgery did little to improve blindness once this level of damage to the optic nerve had been reached (9). The use of corticosteroids to reduce swelling of the optic nerve prior to surgery or as a non-surgical option has shown some success in vision preservation, but this has not proven to be of consistent benefit (1,2). Treatment with bisphosphonates to inhibit osteoclastic and osteoblastic activity is another medical option, but it is difficult to measure success other than with symptoms (3,8). Now that the genetic basis for this disease is known, there is hope in treating its root cause.

TEACHING POINT

Fibrous dysplasia can impersonate many of its fellow lytic lesions and is best diagnosed with correlation of clinical, laboratory, histological, and imaging findings. Involvement of the skull base can cause significant functional deficits, such as vision loss in this case.

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indications for decompression. *J Plast Reconstr Surg.* 2007 Dec; 120(7):1957-62.

FIGURES

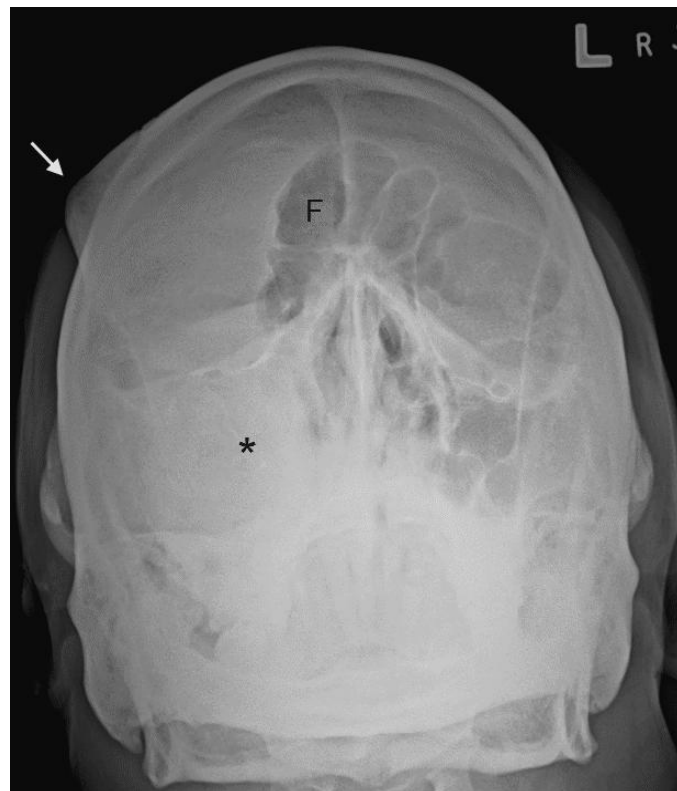


Figure 1: 32 y/o male with craniofacial polyostotic fibrous dysplasia. A submental-vertex view conventional radiograph of the skull demonstrates a triangular-shaped 7mm bony prominence along the right frontal/parietal region (arrow). There is increased density of the right maxillary sinus (asterix). The right frontal sinus is hypoplastic to some degree (F).

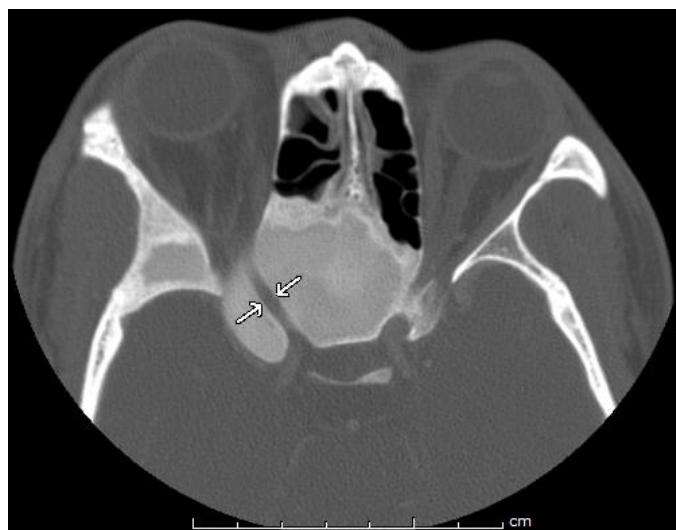


Figure 2: 32 y/o male with craniofacial polyostotic fibrous dysplasia. Contiguous 2.5 mm axial CT imaging was obtained through the facial bones. Bone window utilized for this study. Narrowing of the optic canal is present on the right side which is approximately 3.25 mm in diameter (between arrows). Acquisition was performed using a GE Lightspeed 16 scanner, at 300mA, 120 KVp with 75 ml Omnipaque contrast.

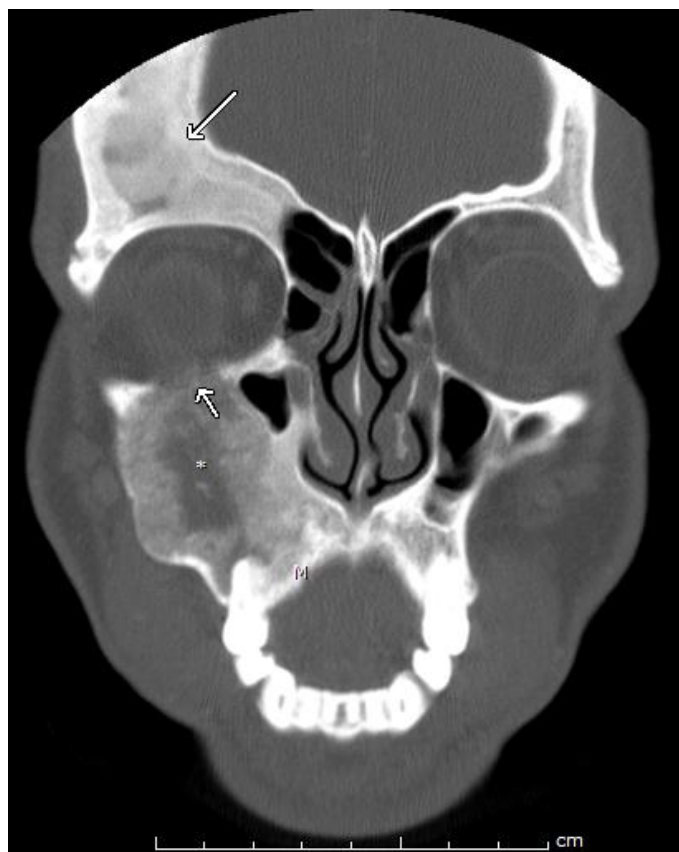


Figure 3: 32 y/o male with craniofacial polyostotic fibrous dysplasia. Contiguous 2.5mm axial CT imaging was obtained through the facial bones. Bone window utilized for this study. A coronal reformatted image reveals complete opacification of the right maxillary sinus (*), extending into the right aspect of the maxilla inferiorly (M), the orbital roof and into the right aspect of the frontal bone in the region of the forehead (long arrow). Disruption of the inferior orbit is noted (short arrow). Acquisition was performed using a GE Lightspeed 16 scanner, at 300mA, 120 KVp with 75 ml Omnipaque contrast.

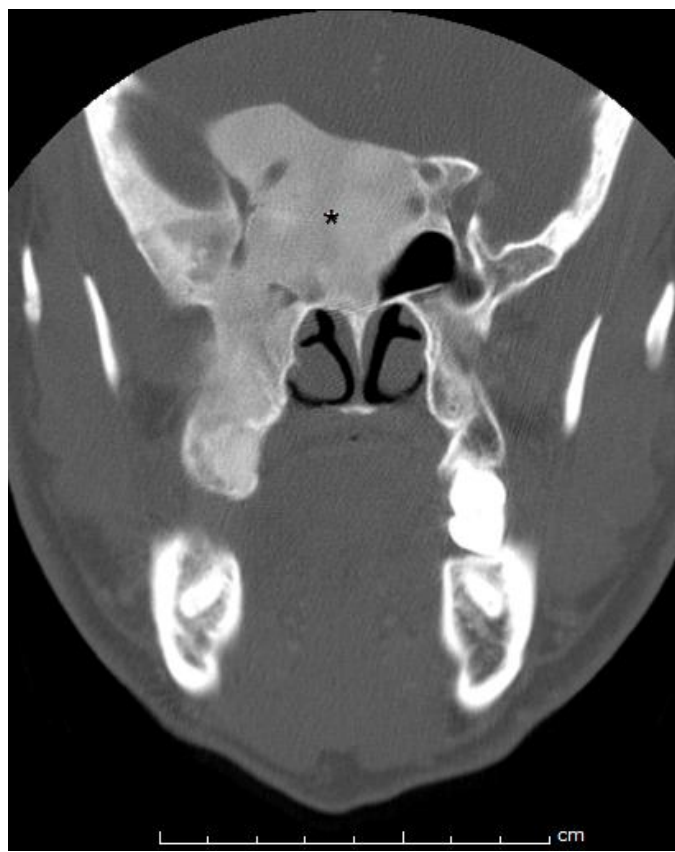


Figure 4: 32 y/o male with craniofacial polyostotic fibrous dysplasia. Contiguous 2.5mm axial CT imaging was obtained through the facial bones supplemented with coronal reformations. Bone window utilized for this study. A coronal reformat demonstrates involvement of the skull base (asterix). Acquisition was performed using a GE Lightspeed 16 scanner, at 300mA, 120 KVp with 75 ml Omnipaque contrast.



Figure 5 (right): 32 y/o male with craniofacial polyostotic fibrous dysplasia status post decompression of right optic nerve. Contiguous 2.5mm axial CT imaging was obtained through the facial bones. The right optic canal measures approximately 7.5 mm following the procedure (between arrows). Acquisition was performed using a GE Lightspeed 16 scanner, at 300mA, 120 KVp with 75 ml Omnipaque contrast.

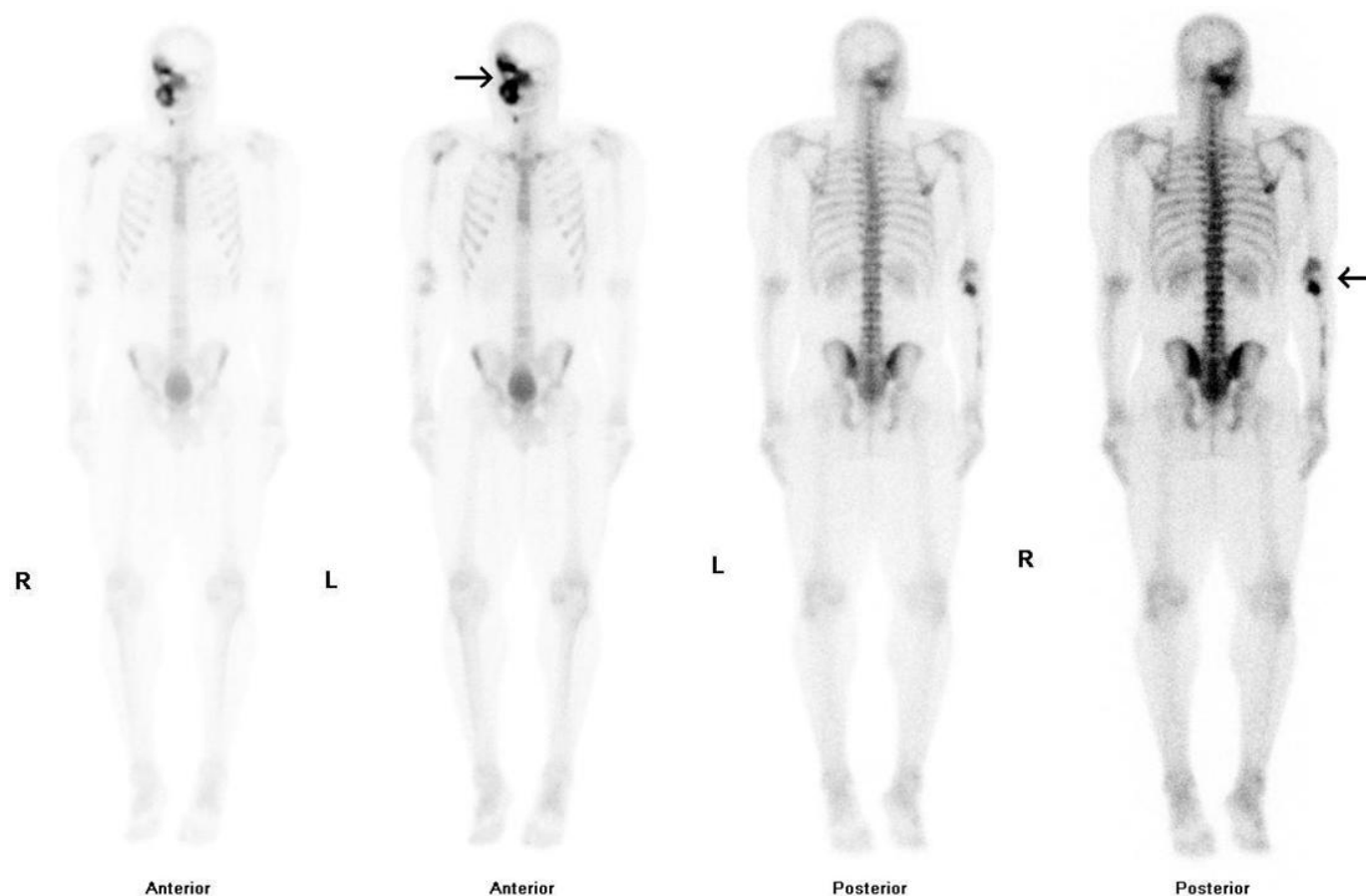


Figure 6: 32 y/o male with craniofacial polyostotic fibrous dysplasia. 20.8 mCi Tc99m MDP were given intravenously, and delayed planar whole body images were performed in the anterior and posterior projections. There is intense radiotracer activity seen about the right head and face region. There are additional foci of activity of MDP activity in the proximal, mid, and distal right humerus. Additional activity is seen in the proximal and mid right forearm (arrows).



Figure 7: 32 y/o male with craniofacial polyostotic fibrous dysplasia. AP (A) and lateral (B) view conventional radiographs of the right humerus with proximal ulna and radius in extension and flexion demonstrate mixed lytic and sclerotic elongated foci without significant bony expansion to involve the proximal, mid, and distal humerus, and proximal mid ulna (asterix).

Etiology	Normal bone replaced by fibroblasts. Mutation in Gs α gene which is located on chromosome 20q13.2-13.3.
Incidence	2-5% of all bone tumors. Polyostotic form 1/6 th as likely.
Gender ratio	No gender predilection
Age Predilection	First decade of life
Risk Factors	None
Treatment	Local excision +/- bisphosphonate
Prognosis	Good, depending on location
Imaging Findings	No periosteal reaction on plain radiographs with early lytic and later sclerotic appearance. Classic “ground-glass” or “smoky matrix” appearance is unreliable. CT or MRI more useful in surgical planning with the variable appearance of fibrous dysplasia.

Table 1: Summary table for Fibrous Dysplasia

Benign Lytic Bone Lesion	Characteristics
Fibrous dysplasia	No periosteal reaction
Enchondroma	1. Calcification 2. Painless
Eosinophilic granuloma	Younger than 30 y/o
Giant cell tumor	1. Epiphysis closed 2. Against articular surface 3. Well defined (nonsclerotic) 4. Eccentric
Nonossifying fibroma	1. Younger than 30 y/o 2. Painless 3. Cortical in origin
Osteoblastoma	Associated with aneurysmal bone cyst (posterior spine)
Metastatic disease	Older than 40 y/o
Aneurysmal bone cyst	1. Expansile 2. Younger than 30 y/o
Solitary bone cyst	1. Central location 2. Younger than 30 y/o
Brown tumor	Other clinical evidence of hyperparathyroidism
Infection	Cannot exclude
Chondroblastoma	1. Younger than 30 y/o 2. Epiphyseal
Chondromyxoid fibroma	No calcified matrix

Table 2: Differential diagnosis for benign lytic bone lesions (4)

ABBREVIATIONS

CT = computed tomography
MRI = magnetic resonance imaging
MDP = methylene-diphosphonate

KEYWORDS

Fibrous Dysplasia, Polyostotic, Craniofacial, Optic Nerve Decompression

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