30-Year-Old Man With Painless Forehead Swelling, Proptosis, and Diplopia

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**History and Chief Complaint:** 30-year-old man with a painless swelling of the left face for many many years. Now, he presents with diplopia and proptosis of the left eye.

**Physical Exam:** Expansile deformity of the skull involving the left forehead and orbit, with proptosis (Figs. 1 and 2).

For the answer, please see the next page.
FIGURE 1. (A and B) There is abnormal swelling from the medial right orbit across to the left lateral skull.

FIGURE 2. (A and B) Enlargement of many of the left-sided facial bones—including the orbital roof and calvaria—with a “ground glass” appearance of the enlarged marrow cavity and diploic space.

Findings: Enlargement of the diploic space with a “ground glass” appearance of many of the left-sided facial bones including the frontal, sphenoid, and nasal bones.

Differential Diagnosis for Bone Thickening/Enlargement:
- Fibrous dysplasia
- Meningioma induced hyperostosis
- Paget disease of bone
- Proteus syndrome
- Metastatic breast or prostate cancer

Case Diagnosis: Fibrous dysplasia
Diagnosed by physical examination with typical imaging appearance on radiographs and computed tomography (CT).

Treatment and follow-up: No immediate treatment needed at this point. The patient will consult for follow-up care and observation.

Discussion: Fibrous dysplasia (FD) is a developmental disorder of bone in which the normal medullary space is replaced by fibro-osseous tissue resulting in expansion of the bone. Four disease patterns are recognized: (1) monostotic FD, most common (75–80%); (2) polyostotic fibrous dysplasia (PFD), often syndromic, comprises 20–25% of cases; (3) craniofacial; and (4) cherubism. Facial bones are involved in up to 25% of all cases, most commonly the maxilla and mandible, but more commonly (up to 50%) in patients with PFD. Pathologic fractures may develop in up to 85% of persons with PFD. Two syndromic associations are recognized: McCune–Albright syndrome and Mazabraud syndrome. Café-au-lait spots are present in one-third to one-half of patients with PFD. McCune–Albright syndrome includes the triad of PFD (usually unilateral), café-au-lait spots (typically on the same side as the bone lesions), and endocrine dysfunction—usually precocious puberty. FD has also been reported in association with endocrine disorders such as hyperthyroidism and hyperparathyroidism. Craniofacial FD typically presents with asymmetric facial swelling—as in this patient. There may be visual symptoms due to encroachment on the orbits, nasal obstruction due to involvement of the nasal fossae, and other cranial neuropathies from expanded bone narrowing their foramina. The maxilla may be severely expanded, distorting facial features—producing the classic leontiasis ossea (“lion face”). Cherubism—an autosomal dominant disorder—causes symmetric involvement of both the maxilla and mandible that typically presents by age 7. FD may increase during pregnancy. Histologically, irregular and deformed bony trabeculae made up of immature woven bone are seen, which are embedded into a fibrous stroma. These irregular trabeculae have been referred to as Chinese characters. This woven bone does not develop into the normal lamellar bone. The normal mechanical integrity of the bone is lost, which is most pronounced in the weight bearing bones in polyostotic FD. The radiologic appearance varies with the degree of fibrous tissue present. The most frequent computed tomography description is that of a “ground glass” appearance. Expansion of the diploic space is seen. On MR imaging, FD is typically of low-to-intermediate signal on all sequences, with intense enhancement post contrast. Rarely FD may undergo malignant transformation (0.5%). The patients will frequently complain of increasing pain, or enlargement of a soft tissue mass. The most common malignancy is osteosarcoma, followed by fibrosarcoma and chondrosarcoma. Malignant transformation can be seen in both the monostotic and polyostotic forms. One-third of these patients have a history of prior radiation therapy.

SUGGESTED READINGS
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