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Craniofacial fibrous dysplasia

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Abstract
Fibrous dysplasia is a benign localized bone disorder of unknown aetiology in which endocrinopathies, abnormal pigmentation of the skin and mucous membranes may form part of the disease. Five cases involving the facial and cranial bones are described to illustrate the various presentations and radiological appearances including computerized tomography and magnetic resonance imaging. Some of these cases demonstrate that serial radiology may be adequate for diagnosis and management without the necessity for histological confirmation. The place of limited surgical treatment for the craniofacial sites is debated.

Introduction

Fibrous dysplasia as an entity has been described for about 50 years but it's management remains controversial. Trials of therapy in craniofacial disease are difficult to perform due to the small number of cases presenting to each otolaryngology unit and the great variation between presentations. The following cases illustrate the underlying principles of individual patient management.

Case 1

A 20-year-old man presented with a one-year history of recurrent left otitis externa and left hemifacial hypertrophy. On examination he had a left meatal stenosis and a conductive hearing impairment of 60 dBHL. This ear was explored and a bony mass was curetted easily from the inferior and posterior canal walls. The tympanic membrane was intact and the annulus was preserved. Histology demonstrated superficial woven bone and deeper lamellar bone. Post-operatively the meatus remained patent and there was resolution of his conductive hearing loss. Six years later, he developed a further lesion in the mandible causing deformity and this was debaulked (Fig. 1).

Case 2

A 28-year-old man presented with a two-month history of right otalgia and hearing loss. His meatus was slit-like due to a bony mass based anteriorly, causing a 50 dBHL conductive hearing impairment. Although there was no facial deformity, the radiological features of fibrous dysplasia were seen in the right maxilla, sphenoid bone and squamous termporal bone. Hypocycloidal tomography demonstrated involvement of both the external and internal auditory meatus (Fig. 2). An endaural approach with an anterior tympanomeatal flap was used to expose the anteriorly based bony mass which was easily curetted to reveal a normal tympanic membrane. The newly excavated external meatus was maintained by BIPP pack for three weeks. Histology revealed the typical features of fibrous dysplasia. The patient remains asymptomatic four years after operation.

Case 3

A 37-year-old builder presented with left nasal obstruction, occasional facial pain but no diplopia, anosmia or epistaxis. There was a large nasal polyp filling the left nasal cavity. In addition, he was noted to have frontal bone asymmetry with displacement of the left orbit but no ophthalmoplegia (Fig. 3). Computerized tomography revealed a mass of dysplastic bone involving the medial wall of the left maxillary antrum, ethmoid sinuses, most of the sphenoid bone and sinuses. The frontal

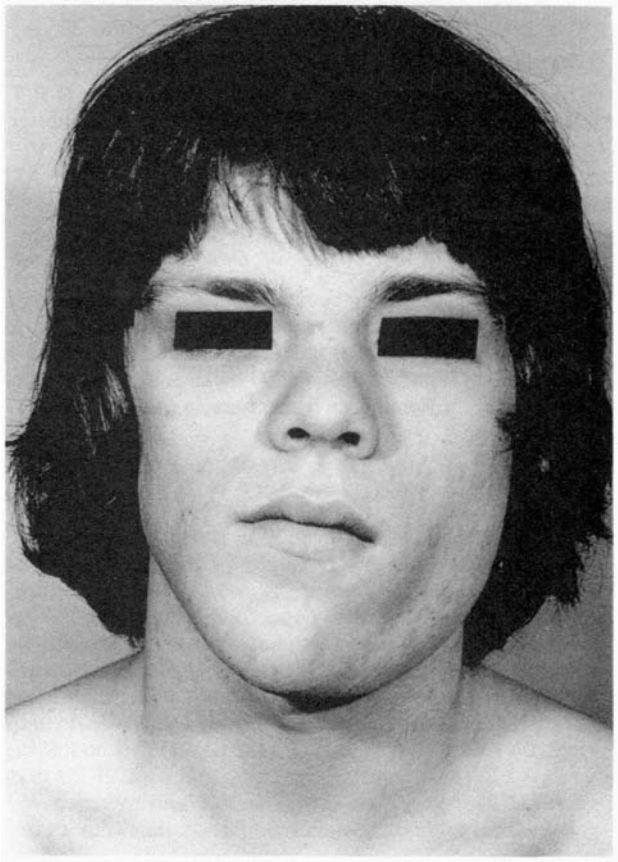

Fig. 1
Hemi-mandibular hypertrophy due to fibrous dysplasia in a 20-year-old man.

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FIG. 2
Hypocycloidal tomography from a 28-year-old man indicating fibrous dysplasia in the whole of the right petrous temporal bone but sparing the labyrinthine capsule.

bone was involved displacing the orbit (Fig. 4). Intranasal polypectomy and incisional biopsy of the bony mass through a Howarth's incision was carried out. The polyp histology was reported as inflammatory tissue and the bony mass was found to consist of bony trabeculae of irregular size and shape composed mainly of woven bone with no surrounding osteoblasts. The stroma was fibrous in appearance. No further treatment was offered to this patient as he had no orbital complications and was not worried about his facial deformity. He is being reviewed at yearly intervals and has been warned to return if there is any change in his vision.

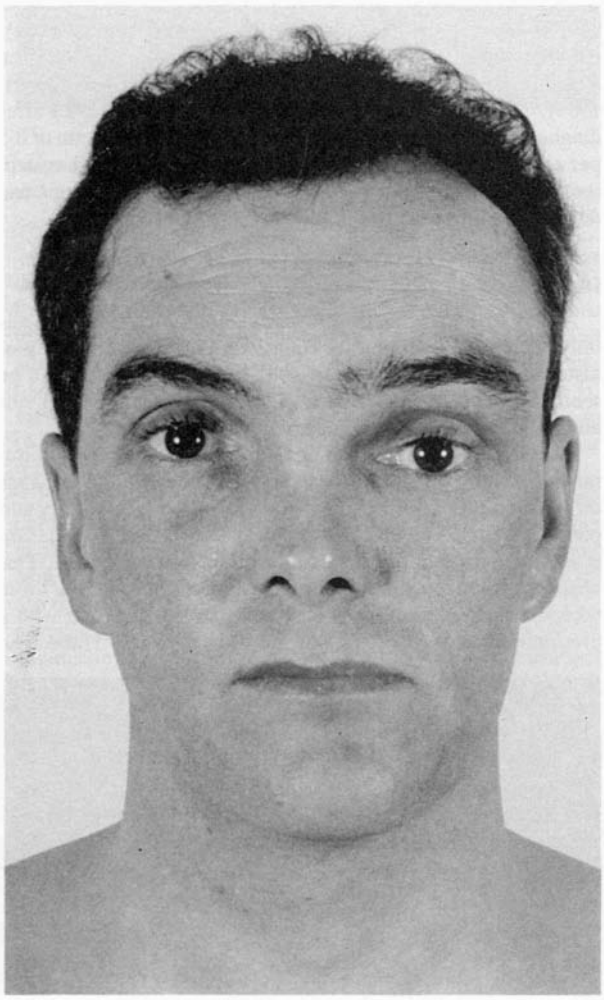

FIG. 3
Displacement of the left orbit by fibrous dysplasia.

Case 4

A 35-year-old lady presented with a unilateral sensorineural hearing loss and diplopia due to an ipsilateral sixth cranial nerve palsy. Plain X-ray of the internal auditory meatus revealed expansion of the left petrous apex by sclerotic bone of fibrous dysplasia. She has since been lost to follow-up.

Case 5

During an episode of conjunctivitis, a 20-year-old woman was noted to have slight infero-lateral displacement of her right orbit without diplopia. Radiological investigation revealed a fibro-osseous lesion involving the whole of the roof of the right orbit but not narrowing the superior orbital fissure or optic canal. She was reviewed, annually, for six years without radiographic change in the size of the lesion which is typical of fibrous dysplasia (Fig. 6).

Review

Prior to the 1930's, cystic bone disease was ascribed to parathyroid disease despite surgery failing to demonstrate a parathyroid adenoma or primary hyperplasia. Recognition of fibrous dysplasia as a specific disease entity began with a report by McCune and Bruch (1937) of a case of osteodystrophica fibrosa in a young female associated with 'café-au-lait' skin pigmentation and precocious puberty. Albright *et al.*, (1937) described a similar case in the same year and the syndrome is now referred to as McCune-Albright Syndrome. Lichtenstein (1938) first suggested the term 'fibrous dysplasia' and pointed out that skin and endocrine changes occurred in only 3 per cent of cases. A hereditary form of fibrous dysplasia that affects the jaws is known as cherubism.

Aetiology

The aetiology of this condition is not clear and several theories exist:

(1) Lichtenstein and Jaffe (1942) proposed that the disease represents a 'congenital anomaly of development' with defective activity of bone-forming mesenchyme, similar to the flaws resulting in hamartomas. Certainly the average age of onset of symptomatology is young (17 years).
(2) Schlumberger (1946), noting that proliferation of connective tissue is a basic response to body trauma, believed fibrous dysplasia to be a reparative process.
(3) Hormonal imbalance has been suggested to explain com-

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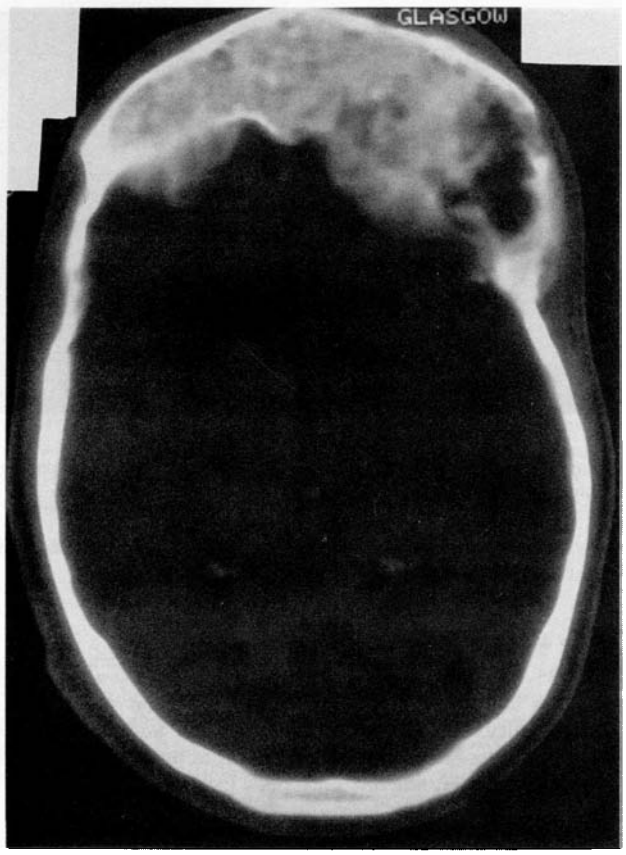

FIG. 4
Computerized axial tomogram from a 37-year-old man with fibrous dysplasia involving the frontal bone bilaterally, obliterating the left frontal sinus and displacing the frontal lobes of the brain.

bined skeletal lesions and precocious puberty (Albright *et al.*, 1937).

(4) Changus (1957), using histochemical techniques, showed that the stromal fibroblasts had high alkaline phosphatase activity and thus were actually osteoblasts. He concluded that fibrous dysplasia represented a hyperplasia of osteoblasts in response to an unidentified stimulus.

(5) Shapiro (Edgerton *et al.*, 1985) believes that fibrous dysplasia results form a 'mutant gene whose protein product affects bone, pigment and hormone metabolism'. It is rarely familial, arises in normal bone and therefore it may result from a mutation of a somatic cell rather than a germ cell.

Clinical Presentation

Fibrous dysplasia is essentially a disease of the young although often going unnoticed until middle-age. The population prevalence has not been reported to this author's knowledge and it is most certainly rare. The bone disease may be monostotic (affecting a single bone) or polyostotic (affecting many bones) and is usually asymmetrical and often unilateral. The rib, femur, tibia and maxilla are most commonly involved. Craniofacial involvement occurs in all of the severe polyostotic forms but in about 30 per cent of the monostotic forms (Fries, 1957). Temporal bone involvement occurs in 18 per cent of craniofacial disease. Apart from the McCune-Albright syndrome, which occurs almost always in females, the sex incidence is predominantly male.

Craniofacial involvement is characterized by a barely noticeable, gradually increasing, painless swelling. It may only be detected at examination and tends to be unilateral. In some instances, vast areas of the skull may be involved (Viljoen *et al.*, 1988). Visual disturbance results from extra-ocular muscle palsies, compression of the optic nerve or chiasm. Paranasal sinus obstruction can cause mucocoele formation but facial pain is very unusual. Excessive tissue vascularity in these lesions can cause symptomatic multiple arterio-venous fistulae.

In the temporal bone, the most common presenting symptoms, in order of decreasing frequency, are progressive unilateral conductive hearing loss, temporal deformity, meatal stenosis, sensorineural hearing loss, and vertigo. Otalgia is rare unless due to a complicating otitis externa. Formation of external meatus cholesteatoma secondary to a severely narrowed canal occurs in one-third of cases. Pre-auricular deformation of the temporal bone has led to temporomandibular joint dysfunction. Facial palsy has not been described, despite radiographic reports of alteration of the Fallopian canal. Mono/polycranial neuropathy and Eustachian tube dysfunction have also been described (Fernandez and Calvita, 1980).

Sarcomatous transformation has not been reported in the temporal bone as it has in other sites (Schwartz and Alpert, 1964). The mean interval of development of malignancy after diagnosis of fibrous dysplasia was 13.5 years giving a rate of 0.5 per cent which compares with 4 per cent for Paget's disease in untreated cases. An increasing growth rate, evidence of bone destruction and pain would indicate such a process.

Diagnosis

As no one investigation is pathognomonic, the diagnosis only becomes apparent after collation of history, examination, histology and radiology (Harrison, 1984). However, it can be seen from two of our cases (No. 3 & 5) that a biopsy is not always mandatory for management of the patient.

Histologically, it is characterized by an arrest of bone maturation, but the histology does vary considerably and depends on the amount of fibrosis, new bone formation, cellularity and pleomorphism. The basic histological component of the lesion is an overgrowth of fibrous tissue within cancellous bone. This fibrous stroma may have abundant spindle cells in a loose whorled arrangement, or it may be relatively acellular with a predominance of a collagenous ground substance. Trabeculae

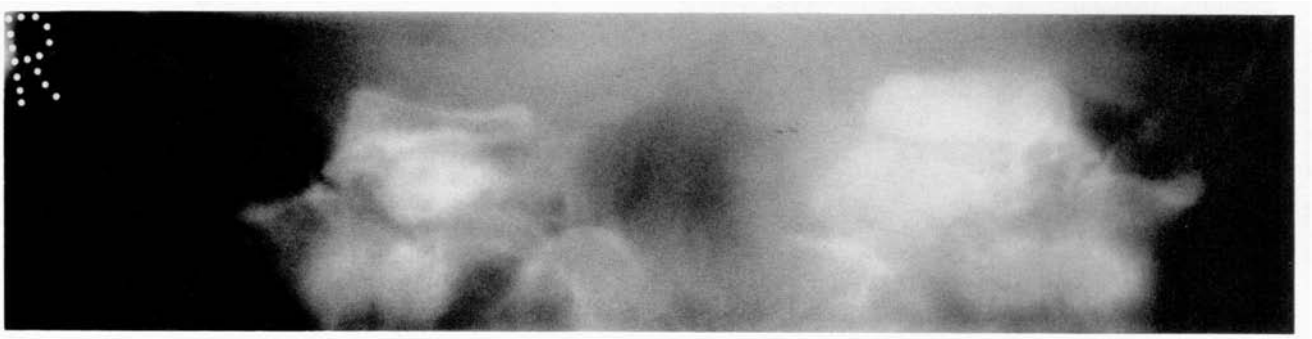

FIG. 5
Isolated fibrous dysplasia of the left petrous bone on hypocycloidal tomography in a 35-year-old woman.

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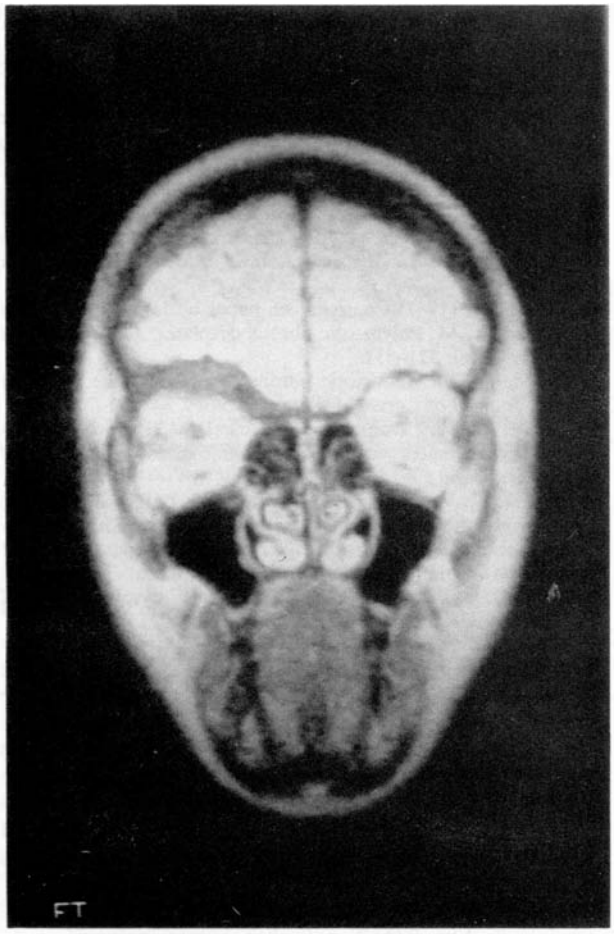

FIG. 6
Magnetic resonance imaging showing the typical cystic bone appearance of fibrous dysplasia in the frontal bone in a 25-year-old woman.

and masses of poorly formed membranous bone are irregularly dispersed with fibrous tissue. Occasionally there are foci of hyaline cartilage and nests of multinucleate giant cells adjacent to areas of blood extravasation. The lesion is not encapsulated and it is impossible to be sure of the true limits of the disease. It has been postulated that in older lesions, lamellar bone (i.e. mature bone) does form which may resemble a benign osteoma (Kwee, 1964). In the frontal bone, fibrous dysplasia can resemble a meningioma.

Fries (1957) has described three radiological patterns in craniofacial fibrous dysplasia. The first is pagetoid with bone expansion and alternate areas of radiodensity and radiolucency. It occurs in more than half the patients, most of whom are older than 30 years of age and have had symptoms for an average of 15 years. The second pattern is sclerotic, with bone expansion and a homogeneous radiodensity (a ground-glass appearance). The third type is cyst-like, usually a round or oval lesion with a sclerotic border. The sclerotic and cyst-like patterns occurred in younger individuals (average age 20 years) with less than three years of symptoms. This suggests that the pagetoid pattern may represent the natural progression of the sclerotic and cyst-like lesions. The ground-glass appearance with an expanded cortex is the most common. The natural history of the disease has two phases: an 'active' phase until puberty and a later 'quiescent' phase. There are, however, many exceptions to this. The most effective method of monitoring growth and estimating extent of disease seems to be computerized tomography.

The serum levels of calcium, phosphate, alkaline, phosphatase, and parathormone are usually all normal. However, severe forms of fibrous dysplasia can have associated hyperphosphataemia. The differential diagnosis in monostotic craniofacial fibrous dysplasia includes osteoma, ossifying fibroma, meningioma, osteoblastoma and osteo/chondrosarcoma. Polyostotic fibrous dysplasia must be differentiated from hyperparathyroidism, Paget's disease, neurofibromatosis and tuberous sclerosis.

Treatment

Chemotherapy has proved ineffective (Williams and Thomas, 1975) in retarding the progression of disease. Oral aluminium acetate can be used to reduce the danger of hyperphosphataemia in severe forms. Steroids have been used with partial success in treating painful lesions. Radiotherapy is contraindicated and has been associated with sarcomatous change (Mock and Rosen, 1986; Sharp, 1970; Schwartz and Alpert, 1964).

Surgery has been recommended in the temporal bone only for reconstruction of a permanently patent external auditory meatus and preservation of cochlear and vestibular function. Excision of all the affected bone is usually fruitless since it is impossible to be sure of the limits of the disease (Nager *et al.*, 1982). If surgery is needed within the 'active' phase, bleeding can be vigorous and regrowth can occur quite quickly. Restenosis of the auditory canal can be prevented by placement of a permanent silastic stent (Sataloff *et al.*, 1985). The patient must also be aware that more than one surgical procedure may be required. Computerized tomography is a significant advance in the therapeutic approach to such cases as it can be used periodically to note any progression and to accurately assess any secondary pathology medial to a stenotic ear canal. In this manner surgical intervention can be more prudently advised and repeated surgery avoided.

Surgery for cranio-maxillo-facial lesions is more controversial. Traditional procrastination during the 'active' phase in children is now increasingly considered unacceptable as there is no indication that surgical procedures inhibit the growth rate of residual normal tissues (Van Horn *et al.*, 1963). Surgery is indicated at any age if important function is threatened, deformity becomes substantial or complications develop *e.g.* obstruction and infection of the paranasal sinuses, dental malocclusion or severe epistaxis. Jackson *et al.*, (1982) argue for total excision of lesions in the cranio-orbital region with iliac or rib graft reconstruction. This provides the only chance of permanent cure as partial removal is associated with a 25 per cent incidence of local recurrence. Unfortunately, total excision frequently results in deformity and functional loss more damaging than the disease itself. Certain features of conservative surgery have proved encouraging. Autogenous bone grafts will successfully unite with remaining bone that still contains elements of fibrous dysplasia. Resected dysplastic bone can be contoured, thinned and replaced as a free graft to aid reconstruction without the risk of apparent regrowth (Edgerton *et al.*, 1985). For this reason, Edgerton suggests conservative surgery with occasional revision for recurrence.

In conclusion, many of these cases can be adequately diagnosed by sequential radiological monitoring and managed by limited surgery for symptomatic complications.

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