Fibrous dysplasia of the orbit

K Bibby, R McFadzean

Abstract

Twelve patients with fibrous dysplasia of the orbit are reviewed and the ophthalmic findings described. Three case histories are presented in detail. Six patients were managed conservatively; four have shown radiological progression of the disease. Six patients underwent surgery. A conservative procedure, comprising debulking dysplastic bone, was carried out in four – all required further surgery including radical excision in two patients. Two subjects had primary radical operations. No recurrence was encountered in the four patients who had undergone radical surgery. It would appear that fibrous dysplasia is not a disease confined to adolescence but may continue into adulthood, and even middle age. Patients may never require surgery, but require follow up for late progression. If surgical intervention is deemed necessary, an attempt should be made to excise all dysplastic bone, since progression of the disease after conservative surgery is relatively common.

(Fr J Ophthalmon 1994; 78: 266-270)

Fibrous dysplasia is a benign, slowly progressive disorder of bone, where normal cancellous bone is replaced by fibrous tissue and immature woven bone. It presents in childhood or early adolescence, typically arresting at puberty.

The lesions were first described by von Recklinghausen in 1891, but it was Lichtenstein in 1938 who recognised the condition as a distinct entity and suggested the name 'fibrous dysplasia'. The condition occurs in the monostotic form, where only one or contiguous bones are involved, or polyostotic form where several distinct areas of the skeleton are affected. Fibrous dysplasia can occur in conjunction with skin pigmentation and endocrine disorders (Albright’s syndrome). The disease has been prevalent for a long time; it was described in the skeleton of a seventh century Anglo Saxon, and in a skull found in Tennessee, dated AD 1480 (plus or minus 130 years, carbon dating).

More than 1500 cases were reported in the literature up until 1971, but estimates of the frequency of fibrous dysplasia of the craniofacial bones vary from 'rare' (5/764 cases of orbital tumours at the Mayo Clinic over 26 years) to 'not uncommon' (144 cases of fibrous dysplasia of the skull).

The aetiology remains obscure, but the condition is widely believed to be a congenital anomaly of bone forming mesenchyme. Hypotheses include misdifferentiation of mesenchymal tissue, an 'arrest of bone maturation in the woven bone stage', and a disturbance of postnatal cancellous bone maintenance. A traumatic aetiology has been suggested but not widely supported.

Seventy per cent of cases involve only one bone/contiguous bones and are classed as monostotic, while 30% of cases are polyostotic. Skull involvement occurs in 10-27% of patients with monostotic, and in 50% of patients with polyostotic disease. Almost 100% of patients with extensive disease have skull involvement. But most patients with affected craniofacial bones have the monostotic form of the disease.

Ocular complications have been classified into primary and secondary processes. Primary complications include involvement of the frontal bone with proptosis; the skull base with extracranial muscle palsies and trigeminal neuralgia; the optic canal with visual loss and optic atrophy; the sphenoid bones with chiasmal compression; and the maxillary bone with epiphora. Secondary complications comprise malignant change, ossifying fibroma formation, and development of a mucocele. Malignant transformation most commonly takes the form of sarcomatous change, occurring spontaneously in 0-5% of cases.

Other tumours known to develop in areas of fibrous dysplasia include cutaneous fibro- myxoma and meningioma.

Materials and methods

Twelve patients with orbital fibrous dysplasia underwent full neuro-ophthalmic assessment, 10 patients presenting for initial diagnosis and two for follow up after management elsewhere. All patients had a skull x ray with serial films to assess progress. Five patients underwent computed tomography (CT) and two patients had magnetic resonance imaging (MRI).

Indications for surgical intervention comprised a deterioration in visual function, disfigurement, and intractable pain. Tissue was sent for histology from all surgically treated patients and a diagnostic biopsy alone was carried out in two patients.

Results

There were five male and seven female patients. Age at diagnosis ranged from 5 years to 45 years,

<table>
<thead>
<tr>
<th>Table 1 Fibrous dysplasia – bones affected</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bone</td>
</tr>
<tr>
<td>---------------</td>
</tr>
<tr>
<td>Frontal</td>
</tr>
<tr>
<td>Sphenoid</td>
</tr>
<tr>
<td>Optic foramen</td>
</tr>
<tr>
<td>Ethmoid</td>
</tr>
<tr>
<td>Maxilla</td>
</tr>
<tr>
<td>Occiput</td>
</tr>
<tr>
<td>Parietal</td>
</tr>
<tr>
<td>Petrous temporal</td>
</tr>
<tr>
<td>Pterygoid</td>
</tr>
<tr>
<td>Mandible</td>
</tr>
</tbody>
</table>
Table 2  Fibrous dysplasia – symptoms

<table>
<thead>
<tr>
<th>Symptom</th>
<th>Number of patients affected</th>
</tr>
</thead>
<tbody>
<tr>
<td>Visual upset</td>
<td>4</td>
</tr>
<tr>
<td>Proptosis</td>
<td>5</td>
</tr>
<tr>
<td>Craniofacial swelling</td>
<td>5</td>
</tr>
<tr>
<td>Headaches/peri orbital pain</td>
<td>5</td>
</tr>
<tr>
<td>Epiphora</td>
<td>1</td>
</tr>
</tbody>
</table>

with an average age of 18 years. Eleven patients had idiopathic fibrous dysplasia, and one had Albright’s syndrome. The skull bones affected are shown in Table 1. Most commonly involved were the frontal bone (eight patients) and the sphenoid bone (seven patients). Bilateral lesions occurred in three patients, one of whom had extensive skeletal involvement. Optic foraminial changes on plain x ray were noted in six patients.

The mean follow up period was 8 years, ranging from 1 to 30 years.

SYMPTOMS

The presenting symptoms are shown in Table 2. Two patients were asymptomatic and diagnosed after routine skull x ray following a trivial head injury, while the patient with Albright’s syndrome presented with increased skin pigmentation and precocious puberty.

Table 3  Ophthalmic findings

<table>
<thead>
<tr>
<th>Patients</th>
<th>Visual acuity</th>
<th>Field loss</th>
<th>Ocular motility defect</th>
<th>Proptosis (mm)</th>
<th>Downward displacement (mm)</th>
<th>Disc</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>CF</td>
<td>Centrococcal scotoma</td>
<td>–</td>
<td>3-5</td>
<td>–</td>
<td>Swollen</td>
</tr>
<tr>
<td>2</td>
<td>NPL</td>
<td>Centrococcal scotoma</td>
<td>–</td>
<td>3-0</td>
<td>–</td>
<td>Pale</td>
</tr>
<tr>
<td>3</td>
<td>6/18</td>
<td>Central scotoma</td>
<td>10° Exotropia*</td>
<td>2-5</td>
<td>–</td>
<td>Normal</td>
</tr>
<tr>
<td>4</td>
<td>6/6</td>
<td>Slight constriction</td>
<td>–</td>
<td>4-0</td>
<td>–</td>
<td>Normal</td>
</tr>
<tr>
<td>5</td>
<td>6/6</td>
<td>–</td>
<td>–</td>
<td>2-0</td>
<td>–</td>
<td>Normal</td>
</tr>
<tr>
<td>6</td>
<td>6/6</td>
<td>Enlarged blind spot</td>
<td>Decreased elevation</td>
<td>10-0</td>
<td>2-0</td>
<td>Swollen</td>
</tr>
<tr>
<td>7</td>
<td>6/6</td>
<td>–</td>
<td>–</td>
<td>2-0</td>
<td>–</td>
<td>Normal</td>
</tr>
<tr>
<td>8</td>
<td>6/6</td>
<td>–</td>
<td>–</td>
<td>2-0</td>
<td>–</td>
<td>Normal</td>
</tr>
<tr>
<td>9</td>
<td>6/6</td>
<td>–</td>
<td>–</td>
<td>2-0</td>
<td>–</td>
<td>Normal</td>
</tr>
<tr>
<td>10</td>
<td>6/6</td>
<td>Temporal constriction</td>
<td>–</td>
<td>2-0</td>
<td>–</td>
<td>Normal</td>
</tr>
<tr>
<td>11</td>
<td>6/6</td>
<td>–</td>
<td>–</td>
<td>3-0</td>
<td>2-0</td>
<td>Normal</td>
</tr>
<tr>
<td>12</td>
<td>6/6</td>
<td>–</td>
<td>–</td>
<td>11-0</td>
<td>–</td>
<td>Pale</td>
</tr>
</tbody>
</table>

CF = counting fingers; NPL = no perception of light.
*This patient had a history of amblyopia.

Table 4  Effects of surgery

<table>
<thead>
<tr>
<th>Patient</th>
<th>Preoperative</th>
<th>Postoperative</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Visual acuity – counting fingers</td>
<td>Visual acuity – 6/60</td>
</tr>
<tr>
<td>2</td>
<td>Proptosis 3-5 mm</td>
<td>Reduced No proptosis</td>
</tr>
<tr>
<td>3</td>
<td>No perception of light</td>
<td>No change 1 mm Proptosis</td>
</tr>
<tr>
<td>4</td>
<td>3 mm Proptosis</td>
<td>Full field</td>
</tr>
<tr>
<td>5</td>
<td>Constricted visual field</td>
<td>Full field</td>
</tr>
<tr>
<td>6</td>
<td>Enlarged blind spot</td>
<td>10 mm Proptosis</td>
</tr>
<tr>
<td>7</td>
<td>Temporal field defect</td>
<td>Reduced 4 mm Proptosis</td>
</tr>
</tbody>
</table>

OPHTHALMIC SIGNS

The presenting ophthalmic signs are shown in Table 3. The commonest sign was proptosis in nine patients, accompanied by downward displacement of the globe in three patients. Loss of visual acuity in three patients ranged from 6/18 to no perception of light.

OTHER FINDINGS

One patient developed conductive deafness secondary to secretory otitis media after blockage of the Eustachian tube.

MANAGEMENT AND OUTCOME

Six patients were managed conservatively, with a mean follow up period of 4-5 years (range 1 to 7 years). Four patients have shown radiological progression without significant change in visual function, while in two patients the disease appears to have arrested clinically and radiologically at the ages of 25 and 29 years.

Six patients underwent surgery. A conservative operation, comprising debulking and reshaping dysplastic bone, was carried out in four patients but all required further surgery, including a radical excision in two patients. Two patients had primary radical surgery.

The surgical and overall outcomes are shown in Tables 4 and 5 respectively. Following surgery, field defects improved in four patients and proptosis was reduced substantially in four patients. However, after conservative surgery all four patients developed recurrent disease, requiring further operations. In one patient the disease remains active after 30 years. No recurrence was encountered in two patients who had radical procedures after conservative operations, or in two patients who had primary radical surgery. The mean follow up period after radical surgery was 3-3 years (range 2 to 7 years).

Postoperative complications developed in three patients, in the form of transient vertical diplopia following frontal bone resection in two patients, and a postoperative subdural haematoma requiring surgical drainage in one patient. There were no long term sequelae.

CASE ILLUSTRATIONS

CASE 1

A 13-year-old boy complained of gradually deteriorating vision in the right eye for 6 months. Visual acuity in the right eye was reduced to counting fingers at 1 metre, with an afferent
pupillary defect and papilloedema. The right visual field showed a dense centrocaecal scotoma and there was 3-5 mm of axial proptosis of the right eye. The left eye appeared healthy.

A CT scan revealed a well defined calcified mass involving the right medial sphenoid (greater wing) and anterior clinoid process, extending around the optic foramen (Fig 1).

A right frontotemporal craniotomy was performed and the dysplastic bone excised completely. Histology confirmed the diagnosis. Visual acuity improved to 6/60 and the centrocaecal scotoma was reduced. The disc swelling and proptosis resolved.

A follow up CT scan showed complete removal of the lesion (Fig 2). This patient has been followed up for 3-5 years with no evidence of recurrence.

Comment
Radical excision resulted in an improvement in visual function and cosmetic appearance in this patient.

CASE 2
A 35-year-old female presented with facial asymmetry, at the age of 5 years. When aged 6 years decompressive surgery to the left maxilla for proptosis and upward displacement of the globe was carried out. Gradual deterioration in the visual acuity in the left eye occurred at the age of 10 years and the left orbit was decompressed. Visual acuity continued to decrease in the left eye until by the age of 14 years she had no perception of light owing to bony occlusion of the optic foramen. At the age of 21 she underwent further cosmetic surgery and when aged 24 years a further decompression of the left orbit was required to reduce proptosis. There was no apparent progression of the disease process during the next 8 years. At the age of 32 years she became pregnant for the first time and during the sixth month of her pregnancy noticed an increase in the size of her left mandible with lumps on her hard palate. Three years later she again became pregnant when the swellings of the left mandible and maxilla further increased in size. A recent skull x ray showed extensive sclerotic bony change involving the left frontal bone, ethmoid, inferior orbital margin, nasal bone, pterygoid plate, maxilla, and mandible (Fig 3). This patient is presently awaiting further surgery.

Comment
Despite five conservative procedures this patient continues to deteriorate both functionally and cosmetically.

CASE 6
A 10-year-old boy developed painless proptosis of the left eye. Initial visual acuity was 6/6 right and left, pupillary reactions and extraocular movements were normal, optic discs were healthy, and visual fields full. A left frontal
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Visual acuity was maintained at 6/6, ocular movements are full, the left optic disc swelling resolved, and the left proptosis was reduced to 4 mm.

Comment
An initial conservative decompression was insufficient to control the functional and cosmetic effects of the disease process. Secondary radical excision has proved beneficial in this patient.

Discussion
In nine of our 12 patients the aetiology of the fibrous dysplasia was unknown (idiopathic), which is the common experience. Two patients attributed their fibrous dysplasia to minor trauma, but it is likely that the association was purely incidental.

The frontal and sphenoid bones were affected most frequently, as in two other series, and the optic foramen was involved in half our patients, a higher proportion than in two other series: 10 of 50 patients and five of 16 patients. This discrepancy may reflect the prolonged follow up of some of our patients.

Three patients demonstrated a decrease in visual acuity – more than in one series, where only two of 16 children had reduced visual acuity (although this may have been because of their young age), but less than in a further series of 10 patients, of whom eight suffered loss of visual acuity. Visual field defects noted in five of our patients are not extensively documented in the literature, although a central scotoma was recorded in one patient and a bitemporal hemianopia in another.

In common with other series, the most frequent ophthalmic sign was proptosis, affecting nine of our 12 patients. Although six patients had radiological evidence of optic foraminal involvement only two developed optic atrophy, and one papilloedema. It is recognised that compressive optic neuropathy does not always supervene, even with anatomical narrowing of the optic canal. Formerly, it was believed that the progression of fibrous dysplasia slowed down or stopped during adolescence. However, many exceptions to this premise are to be found, including five patients in this series whose disease progressed after their teenage years. Only three patients demonstrated spontaneous arrest of their disease, and late reactivation occurred during pregnancy in one patient after 8 quiescent years. Reactivation or exacerbation of active disease is particularly liable to occur during gestation.

Conservative surgery was previously regarded as the treatment of choice. However, the recurrence rate following conservative operations was significant at 25% in two series, and in 20 of 41 patients, 13 of 16, and 13 of 15 patients in other series. Radical surgery was avoided as it was thought that the disease may have a self-limiting course and there was a risk of complete excision causing more deformity and/or functional loss than the disease itself. Following recent advances in anaesthesia and cranio-
facial/plastic surgery, primary radical excision was carried out on five patients without recurrence after 9 years of follow up, while excision of all dysplastic bone in another three patients produced good functional and cosmetic results, although the follow up period was not specified. The four patients who underwent radical operations in our series have demonstrated no recurrence of the disease during an average follow up period of 3-3 years, while all four patients who initially had conservative surgery required further operations. Two patients treated by conservative surgery have required four and five operations respectively and a sixth operation is proposed for the latter.

Four of five patients with visual field loss have improved postoperatively and all have improved cosmetically. Similar results were recorded in two other series and a recent case report. In accordance with the rate of complications for craniofacial surgery in the literature only one of our patients developed a significant postoperative problem in the form of a subdural haematoma, but after surgical drainage there have been no long term sequelae. Attempts to arrest the disease process by radiotherapy in the past have been abandoned owing to an unacceptable 44% malignant transformation rate.

Conclusion
Review of the histories of our 12 patients makes it apparent that fibrous dysplasia is not a disease confined to adolescence but may continue into adulthood and sometimes middle age. Some patients may never require surgery, but do require regular follow up for late progression, particularly during pregnancy. If surgical intervention is deemed necessary, an attempt should be made to excise all dysplastic bone and effect a cure. Progression of the disease after conservative surgery seems to be relatively common.

We thank Mr T A H Hade, consultant neurosurgeon, for referral of these cases for neuro-ophthalmic assessment, Mrs June Cunningham, medical photographer, for the illustrations, and Miss Rachel Gowing for careful preparation of this manuscript.