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Nasal haemophilic pseudotumour

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Abstract
Haemophilic pseudotumour or haemophilic cyst is a rare complication of haemophilia, occurring in one to two per cent of individuals with a severe factor VIII or IX deficiency. We report a case of a haemophilic pseudotumour in a hitherto unreported site, the bony nasal pyramid, and believe this case is also unique on account of it having occurred in a patient with mild haemophilia. The diagnosis and treatment of this rare condition is also reviewed.

Key words: Haemophilia; Nasal bones; Pseudotumour

Introduction

Bone and joint changes secondary to haemophilia were first described by Konig in 1892, but the first documented case of haemophilic pseudotumour was described by Starker in 1918. He described extensive osseous destruction of the femur in association with a massive haematoma in a 14-year-old patient.

This extra-articular involvement of bone associated with a soft tissue mass has been called a haemophilic pseudotumour or a haemophilic bony cyst (Jensen and Putman, 1975). These lesions occur in one to two per cent of individuals with severe factor VIII deficiency (zero to one per cent of normal activity) (Horton *et al.*, 1993) as a result of spontaneous or post-traumatic haemorrhage. If left untreated, these lesions will destroy soft tissue, replace muscle, cause pressure neuropathy and erode through bone. In its end-stage it can erode through blood vessels and skin, causing death by exsanguination or infection. Therefore, early recognition and treatment are required to prevent a threat to life and limb (Gilbert, 1997).

This case report describes what we believe is the first report of a haemophilic pseudotumour of the nasal bones, and possibly only the second case reported in a 'mild' haemophilia A patient (factor VIII activity greater than five per cent of normal).

Case report

This 28-year-old Asian man was referred to the ENT clinic at the Queen Elizabeth Hospital, Birmingham, in April 1997 from the haemophilia unit, where he was on long-term follow-up for mild haemophilia A.

Apart from haemophilia A, his medical history included polyostotic fibrous dysplasia and pulmonary tuberculosis. The former condition was diagnosed when the patient was a child, and on one occasion (in 1983) resulted in intracranial haemorrhage into bony cysts in his cranial vault, requiring a craniotomy for evacuation. He made a full recovery following this procedure. He contracted pulmonary TB following a visit to India in 1987. This infection was successfully treated with a course of anti-tuberculous therapy.

In January 1997 he was reviewed in the haemophilia unit for management of epistaxis following an injury to his nose. He was treated with factor VIII concentration and nasal packing. A plain X-ray of his nasal bones incidentally revealed marked deformity over the bridge of his nose (Figure 1A). The nasal septum was found to be abnormal in texture with some suggestion of a lytic abnormality within it.

The patient was reviewed in the out-patient department three weeks later when the lesion was found to have become bigger. He was thus referred to the ENT clinic. Clinical examination revealed a diffuse but prominent lesion on the dorsum of his nose. Further plain X-rays (Figure 1B) showed a cystic nasal bone lesion, the appearance of which was 'not typical of fibrous dysplasia'. A computed tomography (CT) scan confirmed the presence of an expansile lytic destructive lesion in the nasal bone with an associated soft tissue mass (Figure 2).

Due to the aggressive nature of the lesion a decision was made to debulk the lesion and submit the tissue for histological examination. This was done under a general anaesthetic with adequate factor VIII replacement, through an inter-cartilaginous incision. At surgery, the mass was found to be very fibrotic. The tissue was negative for TB, and histology merely revealed necrotic fragments of fibrous and muscular tissue with areas of old and new haemorrhage and focal stromal haemosiderin pigment. The appearance was not suggestive of fibrous dysplasia. Biopsy was therefore repeated a few weeks later with much the same histological result.

The patient was reviewed after surgery on three occasions and the lesion had become less prominent and had stopped enlarging. He was therefore discharged from further ENT follow-up, although he continues to be seen regularly in the haemophilia unit.

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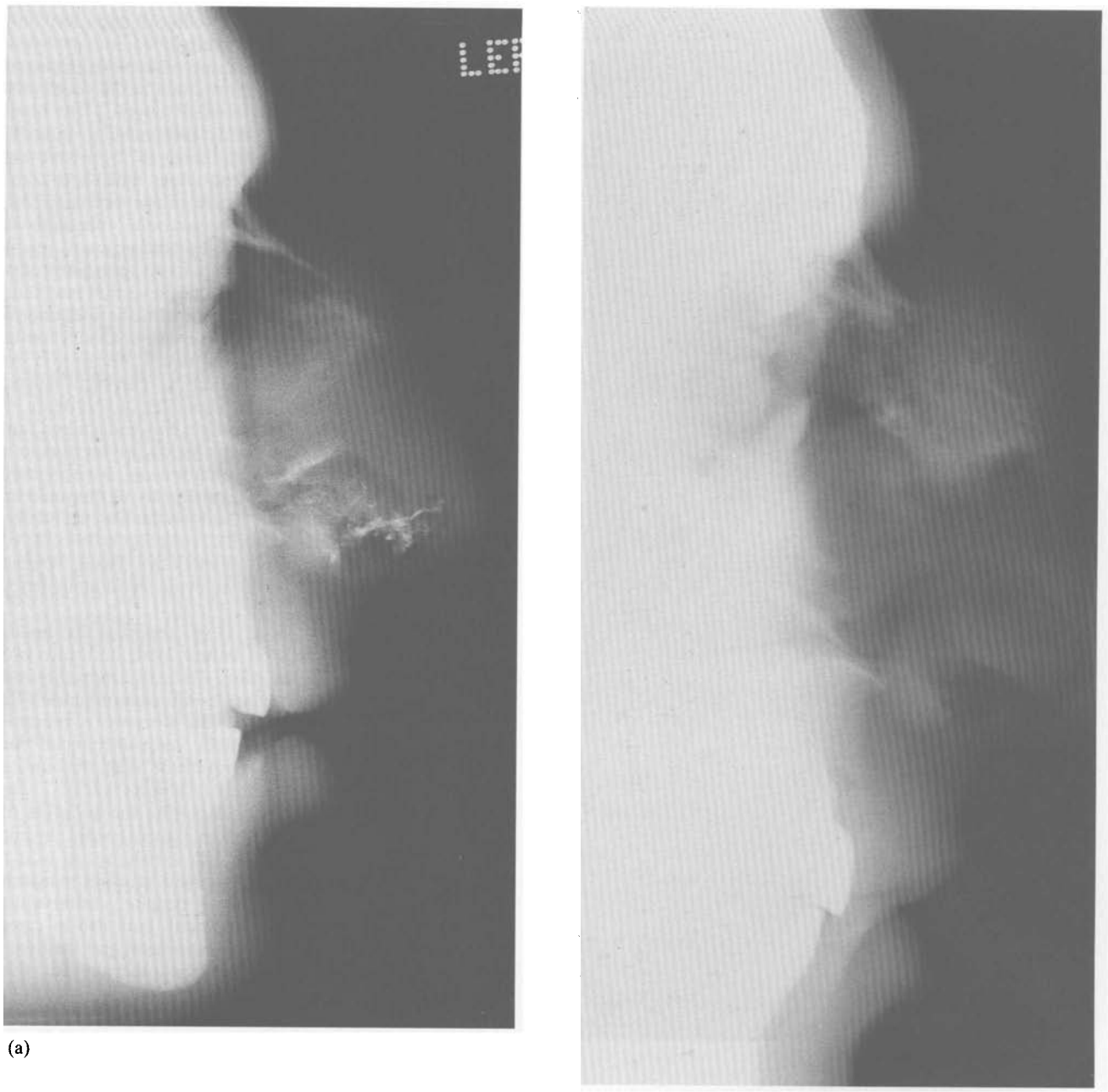

(a) (b)

FIG. 1a and 1b

Lateral plain X-rays of nose taken three months apart showing progressive destruction of the bony nasal pyramid by the haemophilic pseudotumour.

Discussion

Echternacht (1943) credited Starker (1918) with the first description of a haemophilic pseudotumour. These lesions occur most often in the long bones of the lower extremities and the pelvis (Horton *et al.*, 1993). The less common sites of pseudotumour involvement include the orbit (Wenzl *et al.*, 1983; Meyers and Hakami, 1985), the mandible (Stoneman and Beierl, 1975; Mulkey, 1977), the cranial vault (Horton *et al.*, 1993) clavicle, humerus, radius, tibia, calcaneus and small bones of the hands (Jensen and Putman, 1975).

Haemophilic cysts were classified into three major types by Fernandez de Valderrama and Matthews (1965). A Type I cyst is a simple cyst confined solely to the soft tissues within the fascial envelope of a muscle. A Type II cyst originates initially in the soft tissues, but eventually gives rise to changes in adjacent bone due to interference with the local periosteal circulation. The Type III haemophilic pseudotumour is characterized by soft tissue mass in proximity to bone with or without radiological evidence of calcification or ossification within the soft tissue mass, and with evidence of bone erosion and new bone formation.

Gilbert (1975) further distinguished haemophilic pseudotumours into two types based on location. Proximal pseudotumours occur primarily in the pelvis and femur and appear mainly in adults. Distal pseudotumours are located mainly in the hands and feet and occur most commonly in children. These also tend to be multiple and have a better prognosis.

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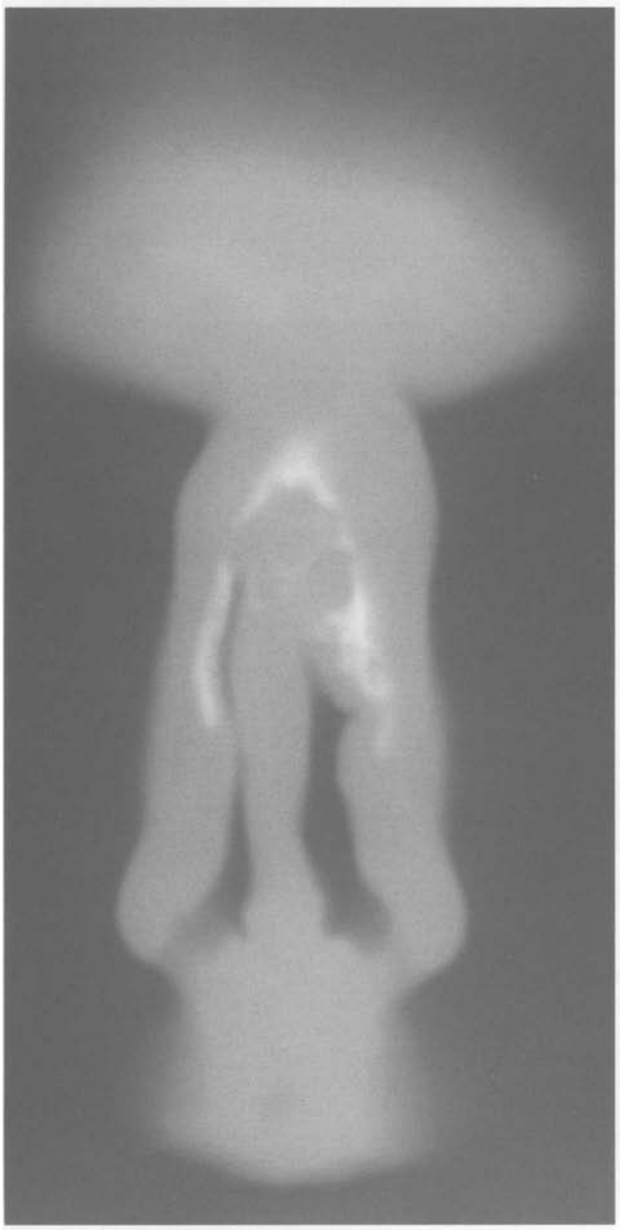

FIG. 2

Coronal CT scan showing an expansile lytic destructive lesion in the bony nasal pyramid with an associated soft tissue mass.

Proposed pathogenetic mechanisms include: (1) extension from a haemarthrosis leading to pressure necrosis and bone destruction (Ghormley and Clegg, 1948); (2) soft tissue or subperiosteal haemorrhage with secondary pressure necrosis and bone destruction followed by new bone formation (Liu *et al.*, 1988) and (3) cortical or medullary haemorrhage followed by cystic changes and late bone destruction and fractures with further haemorrhages.

Given the history of trauma, the location of the lesion and its radiographical appearance, we believe that subperiosteal haemorrhage with secondary pressure necrosis and bone destruction was the operative mechanism in our case.

There is no characteristic histological appearance in these lesions; the usual appearance is of an coagulum with areas of fibrous organization (Liu *et al.*, 1988); there may be clumps of degenerate cells embedded in gelatinous clot (Fernandez de Valderrama and Matthews, 1965).

Diagnosis is usually made on clinical suspicion and radiological appearances. Characteristic radiological features of a soft tissue mass containing coarse calcifications, periosteal elevation, new bone formation in the configuration of bony 'struts' and varying degrees of bone destruction are usually sufficiently distinctive to indicate the diagnosis (Jensen and Putman, 1975). However, roentgenographic appearances on plain films and computed tomography (CT) scans may mimic osteomyelitis, primary bone sarcoma, Ewing's sarcoma, tuberculosis, aneurysmal cysts, echinococcosis (hydatid disease), giant cell tumour, metastatic neoplasm and plasmacytoma (Hussey, 1975). Horton *et al.* (1993) believe that magnetic resonance imaging (MRI) is the diagnostic test of choice because of its sensitivity to blood products. Doubt about the correct diagnosis has led to needle aspiration or biopsies of these lesions resulting in chronic fistulae, infection, further haemorrhages, septicaemia and death.

In our patient, surgical intervention was undertaken due to the rapid increase in size of the lesion (giving rise to a suspicion of malignant transformation in a previously benign lesion), and the history of fibrous dysplasia. The latter diagnosis was excluded based on the absence of the characteristic histological picture of irregular trabeculae of woven bone embedded in a connective tissue stroma (Voytek *et al.*, 1995). There was no evidence of malignancy or tuberculosis.

In a review in 1969, Steel *et al.* reported 16 deaths among the 30 cases managed before 1961, and only two among the 14 cases treated after 1961. The mortality has decreased significantly since the introduction of factor VIII concentrates (Horton *et al.*, 1993). Because a pseudotumour can remain asymptomatic and unchanged for decades, conservative therapy with factor VIII concentrate and immobilization is advocated (Ahlberg, 1975). These measures seem most effective in halting the progress of early lesions but are less useful in patients with chronic lesions (Jensen and Putman, 1975).

Surgical excision has been proposed in cases in which the lesion continues to enlarge despite conservative therapy, or when rupture is imminent, and also in cases of skin necrosis, neurovascular compression and for tissue diagnosis (Horton *et al.*, 1993). Radiotherapy for this condition was first advocated by Muller in 1942. Various doses between 750 and 2000 rads have been used (Liu *et al.*, 1988). It has been recommended especially in patients with factor VIII inhibitors (inactivating antibodies raised against factor VIII), in whom bleeding episodes may be difficult or impossible to control due to lack of efficacy of factor VIII concentrates (Castaneda *et al.*, 1991).

Transcatheter arterial embolization has also been reported for the management of a large iliac pseudotumour (Wessler and Avioli, 1968). Recently, two alternatives to radical surgery have been presented (Gilbert, 1997). Fernandez-Pallazi and Rivas (1985) advocate aspiration of small cysts and the injection of fibrin-glue to obliterate the cavity. This approach is especially suitable in patients with high-responding inhibitors and those with advanced human immunodeficiency virus (HIV) disease.

The introduction of a laparoscope into the cavity of small cysts to evacuate the contents and obliteration of the dead space with bone graft and fibrin-glue has also been advocated.

Conclusion

Haemophilic pseudotumour should be considered in the differential diagnosis of any expanding lesion occurring in patients with severe haemophilia. Conservative management with immobilization and replacement therapy is the

preferred option, with surgery being reserved for tissue diagnosis and when the lesion progresses despite conservative management. Such surgery should only be attempted at centres with experience in the surgical treatment of haemophilic patients.

Our case illustrates the need for a high index of clinical suspicion even in mild haemophilic patients in order to correctly diagnose and treat these unusual lesions.

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