A Case of Pseudotumour in a Haemophiliac
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INTRODUCTION
Pseudotumour is a rare complication of severe haemophilia seen in only 1–2% of persons with the severe form of the disease (1, 2). These lesions were first described by Starker in 1918 as a slow progressive subperiosteal haemorrhage. These lesions may progress relentlessly if left untreated, resulting in compression and pressure necrosis of adjacent structures (1, 3–5). Due to the uncommon occurrence of this lesion, there are no standardized protocols for management. Treatment usually consists of the early and appropriate replacement of factor VIII, and surgical excision of the lesion to prevent further complications if it is progressively enlarging (6).

The case of a young boy with severe haemophilia A who presented to the Orthopaedic Department with a pseudotumour of the left tibia is presented with a review of the clinical and radiological findings.

Keywords: Haemophilia, factor VIII, pseudotumour

CASE REPORT
An eleven-year-old boy known to have haemophilia A (factor VIII < 1%) was transferred to the University Hospital of the West Indies (UHWI) with a two-week history of a painful swelling to his left leg and accompanying fever. There was no history of trauma, but the child had several prior admissions for recurrent haemarthroses of both knees. Of note, his past medical history included an admission to the UHWI some four years previously with a pseudotumour to his right femur. That lesion had been successfully managed by the administration of factor VIII.

The referring hospital reported a proximally swollen, tender, erythematous and warm left leg. No wounds or draining sinuses were noted in the region. There was no effusion to the knee joint, which had a normal active range. No distal neurological or vascular deficits were noted. Blood investigations revealed an elevated erythrocyte sedimentation rate (ESR) of 121 mm/hour, and haemoglobin (Hb) of 7.8 g/dL. Radiographs revealed circumferential erosion of the proximal shaft of the tibia along with narrowing of the proximal fibula.

Blood investigations revealed a haemoglobin of 5.6 gm/dL, white blood cells 6.2 x 10⁹/L, platelets 562 x 10⁹/L, ESR > 130 mm/hour, C-reactive protein (CRP) 5.49 mg/dL, prothrombin time 12.9/12.9 seconds, partial thromboplastin time 106/33 seconds. Factor VIII assay revealed < 1% of native factor VIII activity. Mixing studies performed showed that there was no inhibitor present. He was started on cefuroxime after blood, urine and sputum cultures were obtained. All cultures were sterile. He was placed on strict bed rest and 5 lb of skin traction. Factor VIII replacement was commenced under the guidance of the attending haematologist. The swelling to the patient’s leg continued to expand and was accompanied by pain that was uncontrolled by narcotic analgesics. The leg reached a maximum diameter of 34.5 cm on the third day of admission.

A decision was taken to operatively evacuate his haematoma with recombinant factor VIII to cover the procedure. Prior to surgery, 2500 U of factor VIII (100% correction) was administered, with an additional 2500 U at the time of incision. The aim was to maintain a factor VIII level greater than 70% throughout the procedure (amount of factor VIII is estimated as bodyweight (kg) x 50 = 100%).

A longitudinal incision was made at the anterolateral border of the anterior tibial crest, centred over the tumour. At surgery, a large organized haematoma enveloped by a
pseudocapsule was found and evacuated. The periosteum was elevated by the haematoma and had been stripped off the bone. Samples of clot, periosteum and bone were sent for both culture and histology. There were large cysts in the proximal tibia. The haematoma circumferentially encased the proximal tibia (Figs. 1, 2). Haemostasis was achieved and the wound was closed in layers. In the postoperative period, 1600 U of factor VIII was administered twice daily for three days to maintain levels > 60%.

All intra-operative cultures were sterile with the exception of a sample of the pseudocapsule, which grew coagulase negative Staphylococcus aureus, sensitive to oxacillin and cefuroxime. Histological analysis of his intra-operative samples revealed haematoma.

His postoperative course was complicated by a minor rebleed on day six, three days after his perioperative factor VIII therapy had been discontinued. This minor bleed occurred during attempted non-weight-bearing crutch walking. Factor VIII therapy was recommenced under the guidance of the haematology team, and was continued until the patient’s wound was fully healed. He was limited to mobilization in a wheelchair for the duration of his hospitalization. Cefuroxime was continued until completion of a six-week course, with his CRP returning to normal (< 0.5 mg/dL). He was discharged home in an above knee cast. At six-month follow-up, he was noted to be well with no further episodes of bleeding into the proximal left leg. His left knee was noted to be stiff with loss of the terminal 20 degrees of flexion. No limb length inequality was present.

**DISCUSSION**

A haemophilic pseudotumour is an encapsulated, chronic, slowly expanding haematoma seen in patients with a severe coagulation disorder. Pseudotumours, being a complication of severe haemophilia (< 1% factor), are seen almost exclusively in males aged 20–70 years (7). The index patient was only 11 years old and was already presenting with the second occurrence of this rare phenomenon.

A history of preceding trauma may be associated with the development of pseudotumour in some cases (4, 6). There was no clear recollection of any specific inciting trauma for the index patient. The swelling to the patient’s leg did increase exponentially, however, only after needle aspiration of the proximal tibia was performed at the referring institution.

These lesions usually occur in soft tissues (often intramuscular) but occasionally occur *de novo* in bone or in a subperiosteal location (7). Pseudotumours that occur in muscles with broad tendon insertions readily progress to cause severe pressure erosion of adjacent bone (8). The bones most commonly involved by pseudotumours are the femur, pelvis, tibia, and bones of the hand and hindfoot (7, 9). The index patient had lesions associated with the femur and the tibia which were metachronous.

Pseudotumours are mainly related to the powerful muscle groups of the iliopsoas, quadriceps, triceps, surae and gluteus maximus. All of these muscles have profuse vascular connections with the underlying periosteum and bone, and all have firm connections between their muscle fibres and the periosteum, but not to any great extent with the bone itself.

**Fig. 1:** Anteroposterior radiograph showing erosion of proximal tibia and deformity of fibula (arrow).

**Fig. 2:** Lateral radiograph of proximal tibia with similar findings (arrows).
During growth, the periosteum can be easily detached from the underlying cortex, particularly on the anterior surfaces of the ilium, femur and tibia (4). In the erect position, gravity also acts on all these sites, promoting haemorrhage from damaged vessels.

A pseudotumour consists of clotted blood in various stages of organization and has a fibrous pseudocapsule that contains haemosiderin-laden macrophages (10). As the tumour enlarges progressively, increasing amounts of pressure are exerted on the surrounding structures. In the index case, the proximal tibia was extensively scalloped and the proximal fibula was narrowed and deformed. Bones may be completely destroyed, and muscles and skin are often compressed and may undergo necrosis. Compartment syndromes and joint contractures may also occur (8). There was a flexion contracture of the right knee in the index patient which improved with the evacuation of the haematoma and splinting in extension.

Pseudotumours are usually painless unless complicated by pathological fractures, or nerve compression. Neurologic deficits may occur as a result of nerve compression (6, 7). Superimposed infection or fistula formation to skin or bowel frequently complicates disease management (1, 6). The index case was complicated by a superimposed infection that might have resulted from percutaneous needle aspiration. Profound loss of function secondary to progressive enlargement may occur in the affected extremity (4). In rare instances, exsanguination has resulted from pseudotumour rupture (7, 8). Patients with pseudotumours are often asymptomatic or stable for long periods of time, only to suddenly and unpredictably develop one or more of the complications described earlier (1, 7, 9).

Soft-tissue pseudotumours manifest as nonspecific masses at radiography. They may demonstrate internal calcifications or ossifications. There may be no bone involvement or, at the other end of the spectrum, severe destruction of bone. Subperiosteal bone formation may be present focally and may be the only early radiographic sign of a developing soft-tissue pseudotumour (11). Subperiosteal haemorrhage may cause marked elevation of the periosteum. A chronic pseudotumour on the bone surface may lead to a characteristic curvilinear ossification or calcific “strut” that projects into soft tissues (11). The index patient had a pseudotumour that had eroded the metaphysis of the proximal tibia.

Osseous pseudotumours have a variable radiographic appearance but demonstrate some common features. On radiographs, these lesions are lytic in nature and usually have a well-defined margin; however, they may become quite extensive and completely replace segments of bone (7). Intramedullary and eccentric locations have been noted, and the lesions are often expansile. Osseous trabeculae frequently traverse the lesions. Calcific or ossific foci may be present in the lesions and are presumed to be dystrophic or sequestral in nature (10). There is often reactive sclerosis around bone pseudotumours.

Computed tomography (CT) is useful in determining the osseous anatomy of pseudotumours, while MRI is superior for displaying soft-tissue and intramedullary spaces. Ultrasonography may also help identify the extent of soft-tissue pseudotumours (12).

The MR imaging appearance of pseudotumours is nonspecific but consistent. Invariably, there are heterogeneous low- and high-signal-intensity areas internally on both T1- and T2-weighted images, findings that represent blood products in various stages of evolution (8). A hypo-intense fibrous capsule usually surrounds soft-tissue pseudotumours (13). Acute haemorrhage within a pseudotumour may also be diagnosed on MR imaging. Acute haemorrhage with intracellular deoxyhaemoglobin appears isointense on T1- weighted MR images and hypointense on T2-weighted images (8, 13). The MRI findings of the index case exhibited heterogeneous intensity on T1- and T2-weighted images, with the characteristic hypointense fibrous capsule.

The radiologic differential diagnosis for osseous pseudotumour is largely academic because a diagnosis of pseudotumour can be made confidently when characteristic imaging findings are seen in a patient with a severe coagulation disorder (7). Nevertheless, there are many malignant bone tumours that may resemble a pseudotumour, including fibrosarcoma, plasmacytoma, malignant fibrous histiocytoma, telangiectatic osteosarcoma, and metastatic disease from primary tumours in the kidney, thyroid gland, or lung. Benign bone tumours such as aneurysmal bone cysts, solitary bone cysts, brown tumours, and desmoplastic fibroma are considerations, as well as unusual infectious processes such as echinococcosis. The haemorrhagic nature of osseous and soft-tissue pseudotumours is not helpful in making the diagnosis, as many benign and malignant tumours of the bone and soft tissue with internal haemorrhage have been reported (7).

Due to the high prevalence of complications including life-threatening haemorrhage, fistula formation, and infection, needle biopsy or aspiration of a pseudotumour should never be attempted (1, 6). Indeed, for the index patient, rapid progression of the pseudotumour and superinfection were incited by attempted aspiration of the lesion. Therefore, it is vital that the radiologist make the diagnosis of pseudotumour based on patient history and imaging findings (7).

Therapy for haemophilic pseudotumour should be aimed at maintaining function and minimizing complications. It has been suggested that immobilization and clotting factor replacement works best for recent haemorrhage (14), whereas surgical management yields the best results for pseudotumours that have been present for years or for those in which conservative measures have been ineffective (1, 14). Surgery was indicated for the index patient because of failure of conservative measures. Radiation therapy with doses of 10–20 Gy has been used only in patients who do not respond to conservative treatment and in whom surgery is contraindicated (14, 15) eg due to unavailability of factor VIII or intrapelvic location.
In summary, pseudotumour is a rare complication occurring in patients with severe coagulation abnormalities. A diagnosis should be made based on clinical and radiological findings. Management should be on an individualized basis and requires a collaborative effort between the Haematology and Orthopaedic Departments.

REFERENCES