



Haemophilic Pseudotumor a rare differential for swelling of the great toe A case report. ANAND A

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Abstract : Haemophilic pseudotumor is a rare complication in haemophiliacs with high recurrence rates. Involvement of the bones prone for trauma is common. Early surgical intervention with adequate factor cover proves beneficial in these patients. An 18-year old male with haemophilia A (negative for factor VIII inhibitor assay), presented with a swelling of the left great toe, one year following trauma. It was diagnosed to be a pseudotumor of the left first distal phalanx based on clinical and radiological findings. Amputation of the great toe at the level of first inter-phalangeal joint was performed under factor cover. Post operatively, he was found to have a positive factor VIII inhibitor assay, which warranted increased factor VIII transfusions. 2 years following this, the patient presented with one month history of rapidly increasing swelling of the left great toe. It was a recurrence. 1st ray amputation was performed under factor cover with no post-op complications. This case reinforces that pseudotumor should be considered as one of the differentials for expansile lytic lesions of small bones in the background of haemophilia and suggests close follow up of patients with positive inhibitor assay, considering the high recurrence rate in such patients and the invasiveness of the condition.

Keyword : Haemophilia, Pseudotumor, recurrent pseudotumor, positive factor VIII inhibitor assay

Introduction:

Haemophilic pseudotumor is an uncommon complication in haemophilic patients with clotting factor (VIII or IX) <1% of normal level (moderate to severe haemophilia) and has a reported incidence of 1-2%¹⁻². These occur commonly in bones prone for trauma such as the femur, the ilium, the small bones of the hands and may also occur in lung, intra-abdominal cavity and in the retroperitoneum³. These are cysts that are formed from recurrent bleeding in the extra-articular bones and soft tissues. They present as painless or painful swelling associated with compressive symptoms depending on the region of involvement. There is bone erosion seen on radiographs and it appears dense due to hemosiderin deposits. These features are similar to Ewing's sarcoma, osteo-myelitis, primary osteosarcomas,

metastatic neoplasms, tuberculosis abscesses, giant cell tumors, plasmacytomas and chondrosarcomas. The treatment options include conservative management, minimally invasive procedures such as aspiration, surgical excision and radiotherapy³. The recurrence rate is 15% and is seen most commonly in patients with factor VIII inhibitor⁴. This is thought to occur as a result of hematoma following initial surgery which is prone to occur in patients with Factor VIII inhibitor. Case Report: A 18 year old adolescent male known to have Hemophilia A (Factor VIII inhibitor screen negative) presented to us with swelling of the left great toe for 2 years following trauma, which was insidious in onset, painless and slowly progressive. He gives history of prolonged bleeding after trivial injuries and recurrent episodes of haemarthrosis. There was swelling at the tip of the great toe measuring 4x3cm. It was tender, but there was no local rise in temperature. It was fluctuant, well defined, smooth and fusiform in shape. Plain radiograph revealed an expansile lytic lesion over the left first distal phalanx with cortical breach laterally and the first inter-phalangeal joint was spared. (fig- 1)



figure 1

He was diagnosed to have pseudotumor of the left big toe and he underwent amputation of the left big toe at the level of inter-phalangeal joint, under Factor VIII support. He did not have any undue bleeding during surgery. Microscopic examination of tissue revealed inflammatory granulation tissue with numerous hemosiderophages and areas of old hemorrhage confirming a diagnosis of haemophilic pseudotumor. (fig 2)

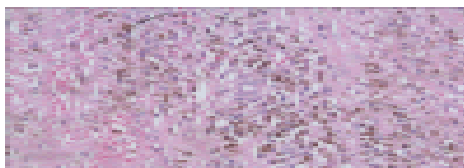


Fig-2 Microscopic examination - confirming haemophilic pseudotumor.

Post operatively he was given continuous low dose Factor VIII (Refacto) infusion for 9 days. On post op day 6, his factor VIII levels did not match the expected levels. Factor VIII inhibitor assay was done which was positive (7 Bethesda units). He was transfused more factor VIII following which the factor level was acceptable and patient had no evidence of bleeding. He was discharged subsequently. 2 years later there was recurrence of swelling which rapidly progressed over one month unlike the initial swelling. This time it measured 9 x 5 cm, extending upto the proximal 1/3rd of first metatarsal of the left foot. The base of first metatarsal and the metatarso-phalangeal joint was palpable. Plain Radiograph revealed a large expansile lesion over the left big toe which has eroded the proximal phalanx and distal half of the first metatarsal bone (fig 3).

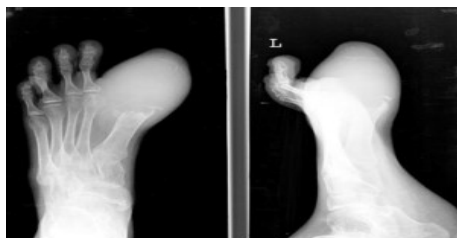


FIG -3

Ray amputation of the 1st toe was performed upto the first metatarsophalangeal joint (fig. 4) under adequate factor VIII cover and antibiotics. There was no intraop or post op bleeding. Wound healing was optimum.

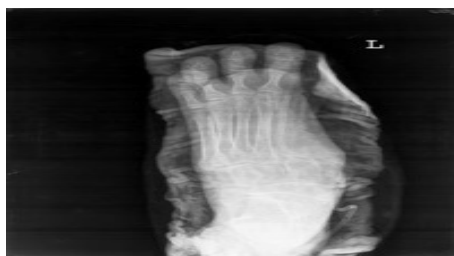


FIG -4

Discussion:

Pseudotumor was first described by Starker in 19185. It usually starts as a muscle hematoma following trauma. It commonly presents as a slow growing, painless cystic mass with variable clinical and radiological features depending on its anatomical location. An osseous pseudotumor mainly occurs in adults and commonly involves the femur, pelvis, tibia and short bones in a descending order of incidence⁶⁻⁷. The development of a pseudotumor results from chronic and recurrent hemorrhages that lead to the formation of an encapsulated mass containing blood at different stages of degradation. Eventually, the hemophilic pseudotumor may progress to a very large lesion. The pseudotumor may develop in soft tissues, or may be intraosseous or subperiosteal, in this decreasing order of frequency. Surgical excision of the pseudotumor is the most effective and curative in

most patients.⁸ With the easy availability of clotting factor preparations, surgical removal of pseudotumors has become the standard modality of treatment. Recurrence rate after surgical excision is 15%⁹ and is seen more often in patients with Factor VIII inhibitor. At radiography, bone hemophilic pseudotumors are seen as osteolytic, well defined expansile, uni- or multilocular lesions. Eventually the lesions show endosteal scalloping, thickening, thinning or even discontinuity of the cortical bone. Marginal sclerosis and dystrophic calcifications may occur. The presence of discontinuous periosteal reaction, which is frequently present in malignant bone lesions, has been described in cases of bone hemophilic pseudotumor¹⁰. In 1972, Brant & Jordan¹⁰ thoroughly reviewed the radiographic findings of pseudotumor, and emphasized the relevance of acute trauma, even when small, related to the hemorrhage that originates the initial lesion. Depending on the number and extent of the subsequent bleedings, this lesion leads to the formation of the pseudotumor that may develop in a matter of weeks or even years. The differential diagnosis for hemophilic pseudotumor may be reached with several benign, malignant or even infectious lesions, especially when presenting intra tumor hemorrhage. At imaging diagnosis, intraosseous hemophilic pseudotumors may simulate several lesions such as giant cell tumors, desmoplastic fibroma, plasmacytoma, metastasis, Ewing's sarcoma, osteosarcoma, aneurysmal bone cyst, brown tumor of hyperparathyroidism and hydatid disease.¹¹ Although several lesions may present imaging findings similar to those of hemophilic pseudotumors, this difficulty is normally overcome by the knowledge of the patient's clinical history. The imaging findings, however, may be specific enough to significantly restrict the differential diagnosis, by demonstrating the capsule of the lesion and the contents with hemoglobin degradation products at different stages. If the pseudotumor is rapidly diagnosed, while the lesion is still small, the clinical management, which includes bone replacement therapy and immobilization, may be resolute. However, in cases where lesion becomes extensive, sometimes involving neurovascular structures, surgery may become the only viable option. Many times, the only surgical option is a mutilating alternative, with amputation of the limb involved¹². MRI may play a significant role in the follow up of pseudotumor progression, particularly in the differentiation between recent bleedings and previous and organized bleedings. Hemophilic pseudotumor very rarely affects great toe and is not reported in literature. However it is a region prone for trauma and therefore in haemophilia patients, pseudotumors can develop in this region following trauma. Hence, it should be considered a differential for swellings of the foot with the back ground of Haemophilia. This patient turned inhibitor positive after the first surgery. These patients are at more risk of bleeding and therefore at more risk of having recurrence. This patient had a recurrence 2 yrs later and the lesion was aggressive. The site of involvement in this patient spared the patient from compressive symptoms. In case of a long bone involvement, the effects of this aggressive lesion could be detrimental. Therefore we suggest close follow up of these patients to pick up recurrence early.

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