Scholars Journal of Applied Medical Sciences (SJAMS) **ISSN 2320-6691 (Online)**
Sch. J. App. Med. Sci., 2015; 3(4B):1707-1711 **ISSN 2347-954X (Print)**
©Scholars Academic and Scientific Publisher
(An International Publisher for Academic and Scientific Resources)
www.saspublishers.com DOI: 10.36347/sjams.2015.v03i04.020

Case Report

Haemophilic Pseudotumor: A Rare Pathological Entity Involving 5th Metacarpal and Associated Tissues

AA Mahmud[1], Y Bashir[2], *MG Taura[3], LH Adamu[3]
[1]Department of Surgery, Aminu Kano Teaching Hospital/Bayero University, Kano, Nigeria
[2]Department of Surgery, Aminu Kano Teaching Hospital, Nigeria
[3]Department of Anatomy, Faculty of Basic Medical Sciences, Bayero University, Kano, Nigeria

***Corresponding author**
Dr. Magaji Garba Taura,
Email: mgtaura.ana@buk.edu.ng

Abstract: Haemophilic pseudotumor is one of the rare complications of haemophilia that results from repetitive bleeding resulting in an encapsulated mass of clotted blood and necrosed tissue. It is uncommon and is seen in severe cases of hemophilia. Complications and symptoms arise due to pain and/or compression of surrounding structures. We present a rare case of a 14 year old boy, haemophilic controlled with factor VIII who presented with a painless right hand swelling, with destruction and resorption of the 5th metacarpal and pressure effect on the surrounding structures. An initial assessment of giant cell tumor was made. The patient had excision and a final diagnosis of organizing haematoma. No haemorrhagic or infectious complications developed after excision.
Keywords: haemophilia, factor VIII, pseudotumor, metacarpal

INTRODUCTION

Haemophilia is an X-linked recessive disorder that occurs because of deficiency of clotting factors. The disease occurs predominantly in males and is transmitted through females. The two common forms of this disorder, hemophilia A and B (due to the deficiency of clotting factors VIII and IX, respectively), are similar in their clinical presentation and imaging findings [1]. Hemophilia A, or the classical presentation of the disease, is more common, with a prevalence of 1:10,000, while haemophilia B or "Christmas disease" is found in 1:50,000 to 1:100,000 individuals [2]. The radiographic features of the rare complication of the haemophilia may mimic many tumor and tumor-like conditions (hence called pseudotumors) and are not diagnostic [1].

Pseudotumors are rare complications of haemophilia, occurring in 1-2% of the patients with severe coagulation disorder [3,4]. It was first described in 1918 by Starker; as an encapsulated, chronic, slowly expanding haematoma with a severe coagulation disorder [5]. Haemophilic pseudotumor of the bone is a rare disease occurring in approximately 1% of patients with severe hemophilia as a long standing complication [6]. It is defined as a progressive cystic swelling by recurrent haemorrhage with roentgenographic evidence of bone involvement [7]. They are categorized as osseous and soft tissue lesions, on the basis of anatomic location [8]. On radiography, intraosseous pseudotumors produce a well-defined, unilobular or multibolar, expansile lytic lesion of variable size. Repetitive bleeding into the bones results in osseous pseudotumor. Osseous haemophilic pseudotumors can be confused with other tumours or infectious condition [8].

Haemophilic pseudotumor usually affects the soft tissue, the long bones of the lower extremities, and the pelvis, and rarely occurs in the orbit, mandible, maxilla, clavicle, tibia, radius, humerus, or small bones [9, 10]. Hence, there are very scanty cases reported in the literature involving the short bones especially the metatarsal bones. The aim of this case presentation was to describe a patient with a haemophilic pseudotumor of right the 5th metacarpal and surrounding tissues and to review the literature on haemophilic pseudotumor.

CASE REPORT

The patient, was a 14 year old male, diagnosed at the age of 2 years following post-uvulectomy bleeding. He was admitted with a swelling on the right hand of four (4) months duration. No associated pain or inability to use the hand. No antecedent history of trauma. Physical examination revealed an oval shaped swelling on medial aspect of the right hand with shiny surface, distended superficial veins, measuring 16cm x10cm, no differential warmth non-tender with a variable consistency ranging from cystic to firm. No limitation of flexion of the fingers.

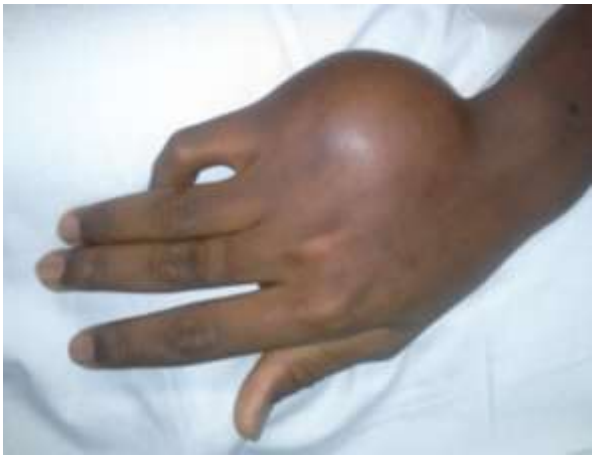

Fig-1: Dorsum of the right hand of a patient presented with haemophilic pseudotumor.

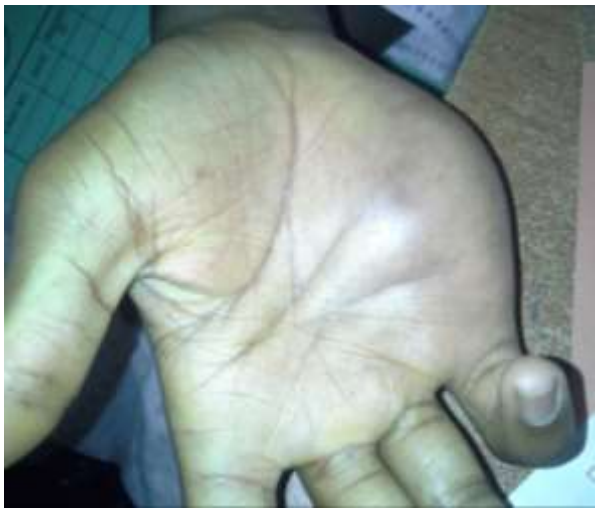

Fig-2: Palm of the right hand of patient presented with haemophilic pseudotumor.

Laboratory tests were as follows; PT (prothrombin time) 15 sec, PTTK > 2min (36-50 secs), INR – 1.00, X-Ray: destruction of the 5th metacarpal with thin rim of whitish demarcation.

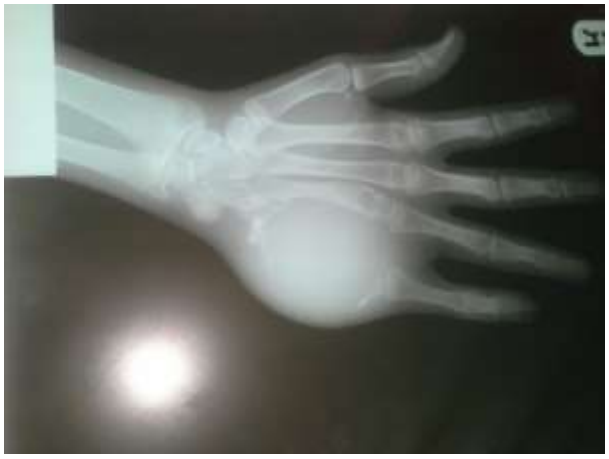

Fig-3: Radiograph of the right hand of a patient presented with haemophilic pseudotumor.

The patient was optimized with adequate amount of factor VIII, placed on 50 iu/Kg factor VIII twice daily, consented for an excisional biopsy which he had. Intra-operatively haematoma was found and fragments of destroyed 5 th metacarpals.

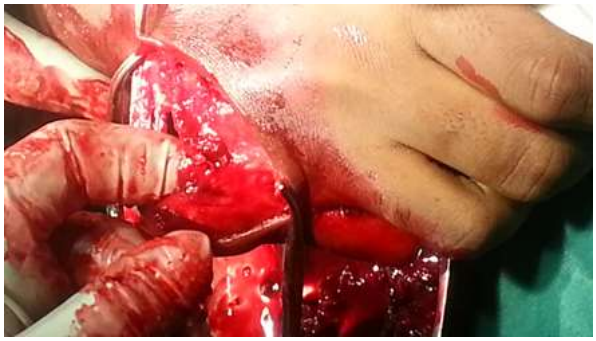

Fig-4: Surgical procedure in the right hand of a patient presented with haemophilic pseudotumor.

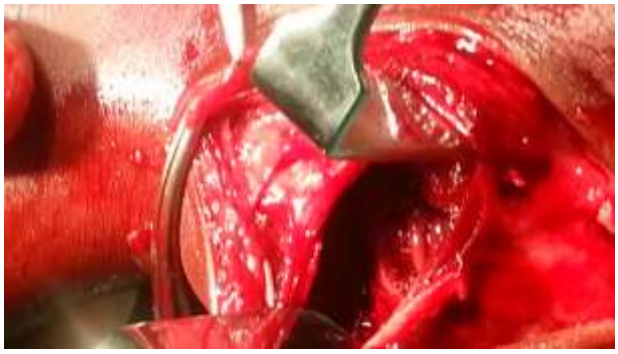

Fig-5: Surgical intervention for correction of haemophilic pseudotumor

Histology showed organizing haematoma. Section showed haematoma collection with areas of gangrene, dystrophic calcification and degenerating red cells. They are surrounded by bony sequestrum and a vascularized pseudocapsule with large blood vessels. Post operatively he was continued on factor viii for a week and discharged.

DISCUSSION

Haemophilic pseudotumor results from multiple episodes of haemorrhage into bones or soft tissue space. Pathological fractures can be associated with intraosseous lesion which can result from bone destruction or resorption due to the chronic pressure of an osseous haemorrhage [10]. Regarding its pathogenesis, it was hypothesized that pseudotumor may be probably due to the pressure necrosis phenomenon, where repeated bleeding into a closed subperiosteal space or intraosseous bleeding areas causes enough pressure to induce necrosis of bone. A new bone being laid down at its edge between the cortex and the displaced periosteum a process referred to as periosteal reaction induced by the haematoma [4]. However, histologically there is no evidence of bone necrosis. Therefore, the causes and processes of formation of this type of pseudotumor continues to be

elusive [11]. It is very clear that the previous case reports on pseudotumors of hemophilia placed emphasis on the location, age of the patient, treatment provided, and outcomes. It was noted that haemophilic pseudotumor usually affects the soft tissue, the long bones of the lower extremities, and the pelvis, but rarely occurs in the orbit, mandible, maxilla, clavicle, tibia, radius, humerus, or small bones, cranium [9, 10, 12, 13, 14]. It was also reported in a region like abdomen [15], among others. Hence, there are very scanty cases reported in the literature involving the short bones especially the metatarsal bones. Here we present a case of 14 year old, male patient with a haemophilic pseudotumor of right 5th Metacarpal bone.

In the present case, the gradually enlarged subcutaneous mass seen oval shaped swelling on medial aspect of the right hand with shiny surface, distended superficial vein also implied haemophilic pseudotumor. However, the differential diagnosis (based on x-ray) could not exclude malignant tumors such as Ewing's sarcoma [16], giant cell tumors [17], chondrosarcoma [18], osteomyelitis [19], tuberculosis [20] and others. Histologic examination was also carried out essentially to establish the diagnosis as recommended by previous authors [14].

For diagnosis of haemophilic pseudotumor of bone, invasive diagnostic techniques such as aspiration are not warranted because severe complication risk is high. Therefore noninvasive techniques should be selected first [6]. In the present case, x-ray was selected as one of the diagnostic techniques. Despite the fact that there are more advanced techniques, the socioeconomic status of the population needs to be put into consideration, as long as desirable outcome will be achieved. Notwithstanding other authors evaluate the advantages of the other imaging techniques, for example MRI is preferred, as it allows recognition of blood products in various stages of evolution. Ultrasonography (USG) shows a central anechoic region with increased echoes behind the lesion due to enclosed fluid in the pseudotumor. Computed tomography (CT) identifies the thick pseudocapsule, but cannot differentiate a haematoma from a chronic abscess [15]. CT is particularly helpful in the evaluation of bone, whereas MRI is superior to CT for delineating soft tissue [21]. Although, various advantages are associated with different techniques, we recommend the use of x-ray as a diagnostic technique.

Current literature indicated that, surgical excision of pseudotumor is the recommended treatment by many authors. However, there are situations where surgical extraction of the condition is not feasible. In such instances, other options including; radiotherapy and arterial embolization should be considered either alone or in adjunction to surgery [22]. Base on the desirable outcome obtained in the present case, we also stress the advantage of the surgical option as one of the treatment modalities. It is noteworthy that the selection of the surgical option may be based on the location of the pseudotumor. It was noted that different complications are related to the location. For instance, the abdominal pseudotumor is associated with complications such as bleeding, bowel perforation and damage to nearby structures due to firm vascular, and nervous adhesions [15]. Although, surgical resection after performing arterial embolization to reduce the vascularization of the pseudotumor is a good alternative, reducing the size of the pseudotumor and the potential risk of bleeding complications during surgery is also important. The time interval will provide room for mass shrinkage but insufficient for vessels restoration [23, 24].

Another case involving the ilium was also aspirated, the procedural complication resulted in the formation of a fistula followed by infection and death from sepsis [4]. Such a fatal course had been reported in the previous case reports. It was agreed that adequate and early substitution therapy for bleeding in muscles and skeleton would prevent the occurrence of pseudotumors [11]. Since 1975, mortality and morbidity has decreased, because treatment of haemophilic pseudotumors has centered on surgical aspirations, surgical removal, supportive therapy in the form of replacement factor, and radiation [25]. Therefore, the most important change has been the use of replacement factor before any surgical intervention [11].

Surgery has been recommended early in management of pseudotumors to prevent erosion of bone, ulceration of skin and infection [7]. Indications for surgery include clinical or radiographic evidence of progressive enlargement of the haematoma, hemodynamic deterioration or occurrence of complication [26]. With index patient, there was complete destruction of 5^{th} metacarpal bone with pressure effect on adjacent bones and soft tissues. There was also resorption of the bone fragments such that only tiny fragment seen intra operatively along with haematoma. The initial diagnosis was based on clinical and x-ray finding and initial assessment of giant cell tumour was made. Invasive diagnostic techniques were avoided to prevent high risk of bleeding. Due to sign of progressive enlargement, surgery was recommended for patients that had being maintained on factor VIII concentrate for a week. The patient was seen at the clinic with no swelling, after 2 weeks stitches were removed.

On the other hand, it is important to note that radiation therapy has been indicated in those cases in which an inhibitor of factor VIII has developed, this result in difficulty to manage the lesion surgically. It was reported that inhibitors to factor VIII have been developed in 15% of patients with factor VIII deficiencies [27]. Other surgical complications that may result from any type of pseudotumor include;

massive haemorrhage, vascular and neurologic damage, and infection, so adequate factor replacement and assessment of any factor that inhibits factor VIII was suggested before treatment [10]. Moreover, coagulate factor levels should be maintained at 100% level for 3 days after the operation, then at 50% in the next 2 weeks [28]. Therefore, proper evaluation needs to be carried out before decision on surgical option as a treatment modality of the case.

CONCLUSION

Haemophilic pseudotumour is a well documented complication of bleeding disorders that clinicians should be aware of while caring for such patients. Signs of compression should be evaluated early because its diagnosis is crucial for evaluation and proper surgical planning, and treatment in conjunction with other specialists. On radiological imaging they mimic other benign or malignant tumors. Haemophilic pseudotumor ought to be one of the top differentials.

Acknowledgements

The authors appreciate the consent given by the parents of the participant as well as those who contributed in the development of this research work.

REFERENCE

1. Jaganathan S, Gamanagatti S, Goyal A; Musculoskeletal Manifestations of Hemophilia: Imaging Features. Curr Probl Diagn Radiol, 2011; 191-97
2. Koenigkam-Santos M, Polezi MB, Engel EE, Pastorello MT, Simão MN, Elias Jr J, Nogueira-Barbosa MH; Uncommon presentations of intraosseous haemophilic pseudotumor in imaging diagnosis. Radiol Bras. 2009; 42(3):159–163.
3. Gunning A, Biggs R, MacFarlane R; Treatment of Hemophilia and Other Coagulation Disorders. Philadelphia, PA: Davis; 1966.
4. Ahlberg AK; On the natural history of haemophilic pseudotumor. J Bone Joint Surg, 1975; 57:1133-6.
5. Liu B, Liu L, Feng Y, Li L; A Case Report on the Surgical Treatment of the Huge Inflammatory Pseudotumor in the AIDS Patient with Haemophilic. Case Reports in Pathol, 2011, Article ID 798649, 4 pages, and doi:10.1155/2011/798649.
6. Kang, JO, Cho, YJ Chul yu, M Hong SE; Haemophilic pseudotumor of the ulna treated with low dose radiation therapy; a case report, Journal of Korean Medical Sciences, 2000, 15; 601-3
7. Meyers L, Hakami N; Pseudotumor of hemophilia in the orbit: the role of radiotherapy in management. Am J Hematol, 1985: 19: 99-104.
8. Resnick D; Diagnosis of bone and joint disorders, 4th ed. Philadelphia: WB Sauders, 2002: 2346-2373
9. Park JS, Ryu KN; Haemophilic pseudotumor involving the musculoskeletal system: spectrum of radiologic findings. AJR Am J Roentgenol, 2004; 183:55- 61.
10. Lewis TR, Webb HR, Bell JP, Beall DP, Fish JR; Haemophilic pseudotumor. J Okla State Med Assoc, 2005; 98:485- 7.
11. Throndson RR, Baker D, Kennedy P, McDaniel K; Pseudotumor of hemophilia in the mandible of a patient with hemophilia A. Oral surgery, oral medicine, oral pathology and oral radiology, 2012;113(2):229-233.
12. Brant EE, Jordan HH; Radiologic aspects of haemophilic pseudotumors in bone. Am J Roentgenol Radium Ther Nucl Med, 1972; 115:525 -39.
13. Hermann G, Gilbert MS, Abdelwahab IF; Hemophilia: evaluation of musculoskeletal involvement with CT, sonography, and MR imaging. Am J Roentgenol, 1992; 158:119- 23.
14. Inoue T, Suzuki Y, Nishimura S, Murakami K, Nishijima M; Cranial haemophilic pseudotumor associated with factor IX deficiency: case report, Surgical Neurol, 2008; 69: 647– 651
15. López-Gómez J, Contreras JS, Figueroa-Ruiz M, Servín-Torres E, Velázquez-García J, Bevia-Pérez F, Delgadillo-Teyer G; Management of the haemophilic pseudotumor of the abdomen: A rarepathological entity. Inter J Surg Case Reports, 2014; 5: 789–792
16. Boumdin H, El Quessar A, Chakir N, El Hassani MR, Jiddane M; Primary Ewing's sarcoma of the cranial vault. Report of 2 cases. Journal of neuroradiology. Journal de neuroradiologie, 2001; 28(3):200-204.
17. Harris AE, Beckner ME, Barnes L, Kassam A, Horowitz M; Giant cell tumor of the skull: a case report and review of the literature. Surg Neurol, 2004; 61:274- 7.
18. Koepke JA, Brower TW; Chondrosarcoma mimicking pseudotumor of hemophilia. Arch Pathol, 1965; 80:655- 9.
19. Riffaud L, Morandi X; Osteomyelitis of the skull vault induced by self-inflicted dermatitis of the scalp. Acta Neurochir (Wien), 2001; 143: 737- 8.
20. Shahat AH, Obaideen AM, Ahmed I, uz Zahman A; Cranial-epidural tuberculosis presenting as a scalp swelling. Surg Neurol, 2004; 61: 464- 7
21. George H, Marvin SG, Abdelwahab IF; Hemophilia: evaluation of muscu-loskeletal involvement with CT, sonography, and MR imaging. Am J Roentgenol, 1992; 158:119–23.
22. Espandar R, Heidari P, Rodriguez-Merchan C; Management of haemophilic pseu-dotumors with special emphasis on radiotherapy and arterial embolization. Haemophilia, 2009;15:448–57.
23. Rodriguez-Merchan EC, Jimenez-Yuste V; The role of selective angiographicembolization of the musculo-skeletal system in haemophilia. Haemophilia, 2009; 15: 864–8.
24. Ahuja SP, Sidonio Jr R, Raj AB, Bertolone SJ, Silverman C, Antekeier DP, et al.; Successful combination therapy of a proximal haemophilic pseudotumourwith surgery, radiation and

embolization in a child with mild haemophilia A. Haemophilia, 2007;13:209–12.
25. Jenson PS, Putnam CE; Haemophilic pseudotumor: diagnosis, treatment, and complications. Am J Dis Child, 1975; 129:717-9.
26. Pakala A, Thomas J, Comp P; Haematophilic Pseudotumor: A case report and review of literature. Inter J Clin Med, 2012; 3: 229-233.
27. Correra A, Buckley J, Roser S, Schreiber A, Syrop S; Radiotherapy of a pseudotumor in a hemophiliac with factor VIII inhibitor.Am J Pediatr Hematol/Oncol, 1984;6:325-7.
28. Rodriguez-Merchan EC; The haemophilic pseudotumour. Haemophilia, 2002; 8:12 - 6