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Hemophilic Pseudotumor in the Leg: A Case Report and Literature Review

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ABSTRACT

Background: One of the less common complications of hemophilia is a hemophilic pseudotumor, which is an encapsulated mass of necrosed tissue and clotted blood that develops from repeated bleeding. These pseudotumor are rarely detected in the leg and are primarily prevalent in the pelvis and mandible.
Case report: A 34-year-old patient with history of hemophilia came with complaints of pain on the left knee to the left calf. Physical examination revealed multiple skin blister on cutis regio cruris sinistra, pain during tibiotalar mobilization, and limited support. Images showed intramuscular hematoma in medial and lateral head gastrocnemius sinistra which forces the left soleus anteriorly. In view of these findings, the patient underwent biopsy with favorable postoperative outcomes.
Discussion: This hematological condition tends to be self-limited and reabsorbed, favoring spontaneous bleeding or minimal trauma. Following the initial hemarthrosis episode, the condition may become chronic or recurring, resulting in gradual cartilage damage and synovial membrane enlargement. Because pseudotumor can occur in places like the leg where prompt identification is impossible, their presentation necessitates a high degree of suspicion. Because there was a lack of soft tissue in the perilesional area, the pseudotumor presentation described here showed early clinical evidence. It is important to note that the most effective treatment is preventing musculoskeletal bleeding with prophylactic coagulation factor delivery.
Conclusion: In order to give appropriate treatment, it is crucial to identify the potential for missing hemophilic pseudotumor at the time of initial diagnosis.

Keywords: hemophilia, pseudotumor, arthrosis, leg

INTRODUCTION

Hemophilia A and B are X-linked clotting diseases caused by lack of factors VIII and IX, respectively. The severity of a disease affects its clinical manifestation. Severe hemophilia causes spontaneous bleeding into joints and muscles. Hemophilia is diagnosed using a combination of a family history, clinical symptoms, and laboratory testing. The primary goal of managing acute bleeding in hemophilia is to induce rapid and vigorous hemostasis, preferably within two hours of the beginning of symptoms, and to rectify coagulopathy. The most serious complication of therapy in hemophilia patients is the development of inhibitors, which are

alloantibodies (IgG) directed against factor VIII and IX that negate its effect.[1] Another serious complication of hemophilia is hemophilic arthropathies, which result from persistent musculoskeletal bleeding. Approximately 90% of severe hemophilia patients who have had multiple musculoskeletal bleeding end up having chronic degenerative abnormalities in key joints such as ankles, knees, and elbows.[2]

One of the less common complications of hemophilia is a hemophilic pseudotumor, which is an encapsulated mass of necrosed tissue and clotted blood that develops from repeated bleeding. Repeated bouts of bleeding usually occur at a bone fracture site, subperiosteal hemorrhage, or bleeding into soft tissues. The prevalence of hemophilic pseudotumor in patients with severe hemophilia is 1-2%.[3] Insufficient resorption of extravasated blood leads to an encapsulated region with clotted blood and necrotic tissue. These lesions develop over time as a result of further hemorrhagic events, finally generating symptoms via mass effect. These pseudotumor are rarely detected in the leg and are primarily prevalent in the pelvis and mandible.[4]

Very few leg pseudotumor have been reported in literature. In this case report, we describe a patient with a hemophilic pseudotumor in the leg and review the literature on hemophilic pseudotumor.

CASE PRESENTATION

A 34-year-old patient with history of hemophilia came with complaints of pain on the left knee to the left calf especially when lying down. The patient's medical history only included hemophilia, which had been diagnosed when the patient was five years old. The patient's psychological condition was normal. The mother was a hemophilia carrier and the father was healthy. According to the medical history, similar complaints have occurred 2 and 3 years ago respectively. Two years ago, patient came with complaints of swelling in the right leg started 1 week, started with a trauma history on the right leg. After the trauma, the leg was getting swollen and darker, sometimes accompanied by blistering and redness. The patient and family does not remember the type of hemophilia and according to the patient it was treated by giving a white liquid in the past.

Based on his current symptom, he noticed numbness in the left upper thigh that were brought on by prolonged weight bearing and relieved by rest. He did not seek medical attention for a substantial period of time. The pain however worsened over time and began to radiate down to his knee. Based on "look" examination in left knee region, it was found swelling around distal thigh until distal foot without bruise or deformity. From "feel" examination, it was found palpable lump at distal femur and proximal tibia with soft consistency, ill-defined margin, with warm and tenderness sensation. On "move" examination, it was found active ROM knee, ankle, and metatarsophalangeal limited due to pain. Upon referral of the patient and on physical examination revealed multiple skin blister on cutis regio cruris sinistra (Figure 1), pain during tibiotalar mobilization, and limited support range of motion.

After examination, taking medical history, and upon primarily suspecting a hemophilic pseudotumor, blood examination with hemophilia factors, pus culture, MRI cruris sinistra, left proximal tibia core biopsy were prescribed. Blood examination found normochromic normocytic anemia, high D-dimer levels, with hemophilia factor VIII and IX within normal limit. Pus culture reveal there was no growth of specific pathogenic bacteria from the examination specimen. MRI images showed intramuscular hematoma in medial and lateral head gastrocnemius sinistra which forces the left soleus anteriorly. In view of these findings, the patient underwent biopsy with favorable

postoperative outcomes. Histomorphology shows a chronic inflammatory depiction with the distribution of hemosiderophage pigments, no neoplastic cells appear in this preparation.

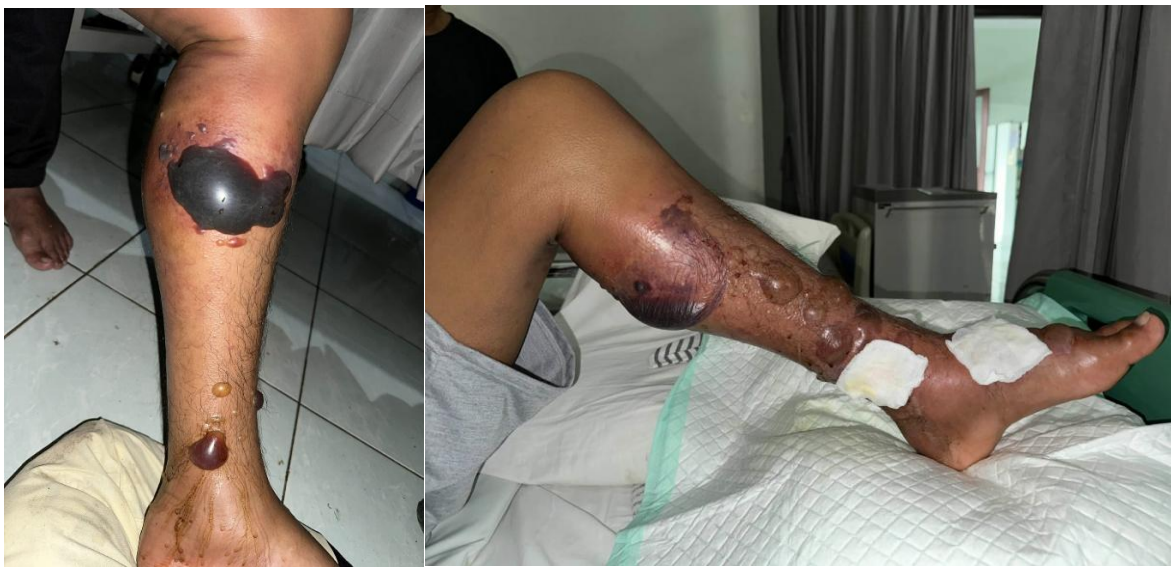

Figure 1. Multiple skin blister on cutis regio cruris sinistra.

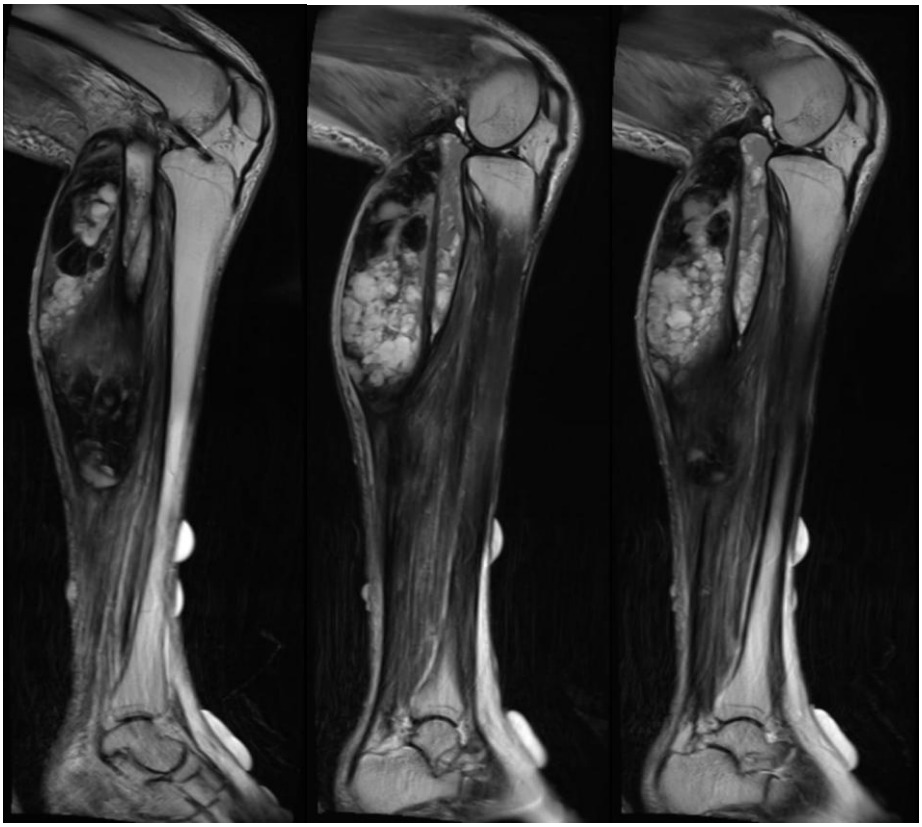

Figure 2. MRI left leg T2W sagittal view.

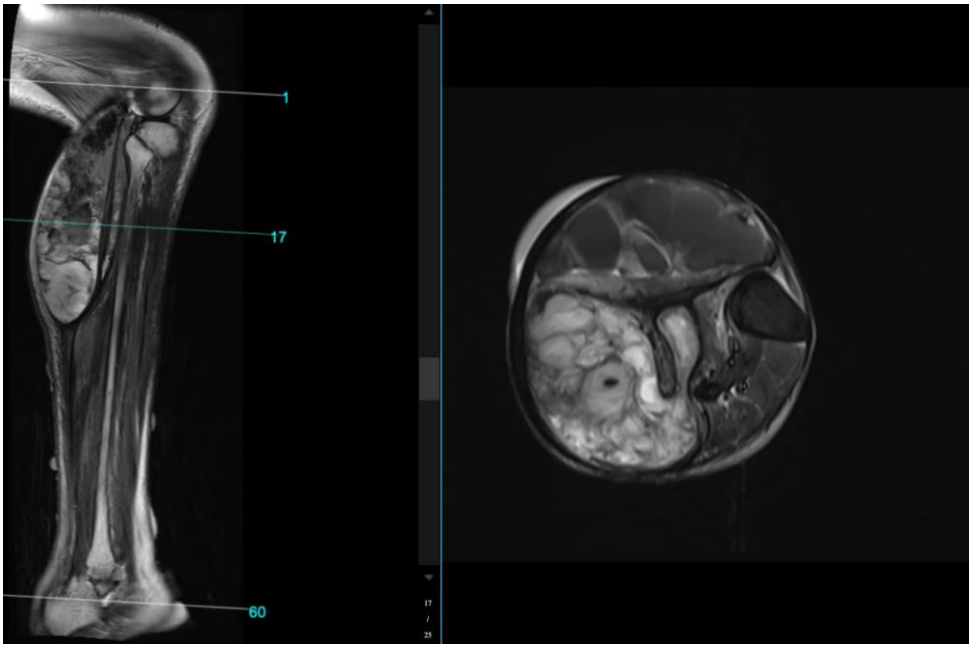

Figure 3. MRI left leg T2W axial view

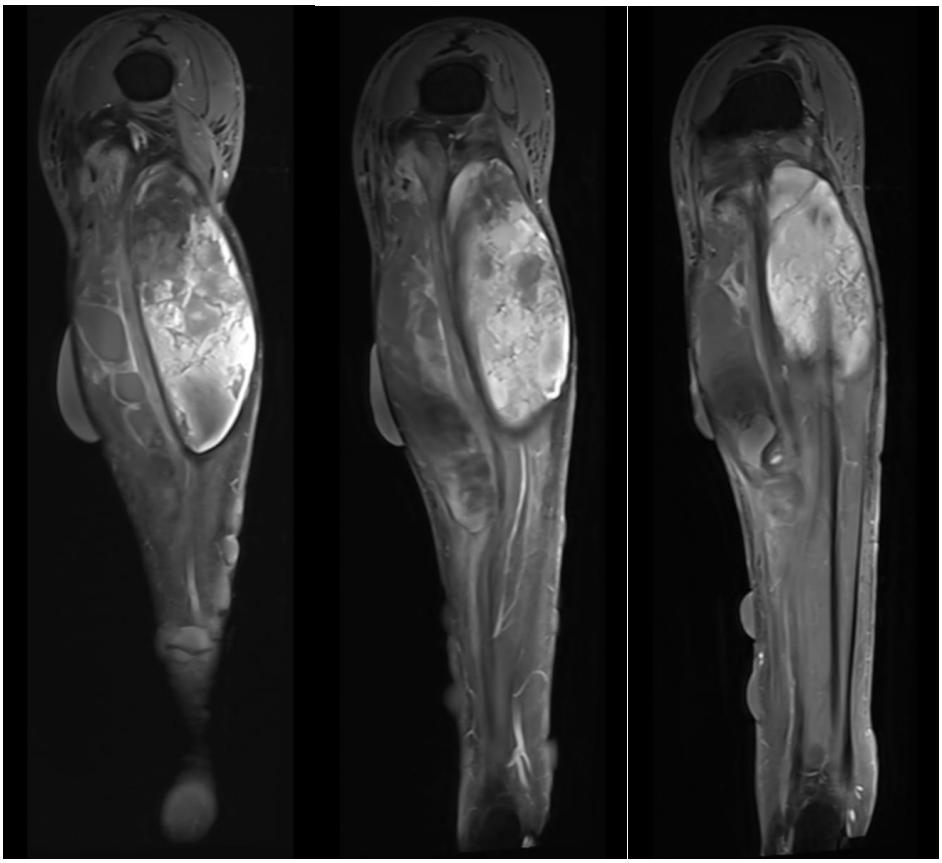

Figure 4. MRI left leg T2W coronal view

There is no consensus on how to treat pseudotumor, thus several options are evaluated based on patient findings and institutional resources. For lesions smaller than 1 cm, aggressive replacement therapy is effective. For larger lesions, administering coagulation factor may help reduce size before surgical excision. At this case, patient was not offered surgical resection of the pseudotumor in the event that his symptoms recurred but weren't deemed to be life threatening.

DISCUSSION

Pseudotumor or "blood cysts" is a rare consequence of severe hemorrhagic syndrome, occurring in about 1% - 2% of patients. Patients with this condition are typically hemophiliacs who have had many hemorrhagic episodes from different sites throughout time.[3] Patients may appear with asymptomatic masses or painful crises caused by abrupt bleeding into the tumor. Pseudotumor cause significant morbidity by compressing adjacent structures, resulting in bone deterioration, muscle necrosis, and skin necrosis. Symptoms vary by area and may include palpable lumps, numbness, weakness, and neuralgia.[5]

The exact pathophysiology of hemophilic pseudotumor still unknown, but several hypotheses have been proposed, including osteonecrosis with hemorrhage, subperiosteal hemorrhage or soft tissue necrosis and bone destruction, and formation of a cystic lesion with bone destruction and hemorrhage.[6] This hematological condition tends to be self-limited and reabsorbed, favoring spontaneous bleeding or minimal trauma. Following the initial hemarthrosis episode, the condition may become chronic or recurring, resulting in gradual cartilage damage and synovial membrane enlargement.[2]

Pseudotumor are made up of extravasated clotted blood enclosed by a fibrous capsule. They typically affect bones, but can also occur in the lung, abdomen, and stomach wall. The bones most typically affected by trauma are the pelvis, tibia, and tiny bones of the hand in decreasing order of frequency.[7] Pseudotumor can occur in places like the leg where prompt identification is impossible, so that their presentation necessitates a high degree of suspicion. Because there was a lack of soft tissue in the perilesional area, the pseudotumor presentation described here showed early clinical evidence. It is important to note that the most effective treatment is preventing musculoskeletal bleeding with prophylactic coagulation factor delivery[8]

Radiologic imaging shows thick hemosiderin deposits within the cyst, which can be mistaken for benign or malignant tumors or infections. MRI is more effective than other imaging modalities for detecting soft tissues. MRI can detect pseudotumor and inform therapy options, as well as monitor tumor response to treatment. Pseudotumor present as a heterogeneous signal on MRI, with a dense fibrous or hemosiderin-laden capsule showing low intensity on T1 and T2 weighted images. Mural nodules are a characteristic feature of intramuscular pseudotumor. The appearance on MRI is non-specific and similar to tumors and abscesses. However, there are heterogeneous low- and high-signal intensity patches on both pulse sequences, indicating the presence of blood products at different phases of evolution.[9,10]

Hemophilic pseudotumor are histologically similar to hematomas and have a dense fibrous capsule. The cyst wall is made of collagenous connective tissue, whereas the cavity contains fibrous tissue, "toothpaste-like" detritus, and liquid clots. The cyst chamber may contain bone fragments, hemosiderin-loaded histiocytes, vascular and osteoid neoformations, gigantic multinucleated cells, and foreign body-type cells.[11] Another unique characteristic for microscopic examination usually indicated that the cyst wall was made up of severely hyalinized fibrous tissue lined on one side by inflammatory granulation tissue containing many hemosiderophages.[12]

Due to the rarity of hemophilic pseudotumor, there is no unanimity among authors on management options. Individualizing therapy is recommended due to variations in techniques among case reports. A conservative therapy involves factor replacement and immobilization of the afflicted area.[13] Long-term replacement

offers a positive prognosis and is recommended for managing pseudotumor induced by recent hemorrhage. Cyst fluid should not be aspirated for diagnostic or therapeutic purposes. First, the cystic contents are too thick for effective aspiration. Second, there is a high risk of recurrence, sepsis, and the formation of a chronic fistula.[7]

Factor replacement therapy has improved hemorrhagic episodes, potentially lowering the risk of pseudotumor. However, factor substitution therapy is not helpful for patients with autoantibodies against factor VIII or factor IX. Bleeding in these patients is typically more severe and occurs in many locations.[8]

Early surgery is suggested for managing pseudotumor of the ilium and femur to minimize bone degradation, skin ulceration, and infection. Surgery is recommended for hematomas with increasing growth, hemodynamic worsening, or complications. Pseudotumor are challenging to surgically remove due to their size, distortion of normal architecture, involvement of surrounding neurovascular pathways, and the presence of daughter cysts that must be removed to avoid recurrence. The primary issue with surgery is the potential of uncontrollable hemorrhage.[14]

When surgery is not an option and conservative treatments are ineffective, radiotherapy can be a viable alternative. Pseudotumor may respond to radiation therapy through multiple ways. Radiation can cause damage to blood vessels and fibrosis in the cellular component of the pseudotumor. Radiotherapy can effectively control recurring long bone pseudotumor.[15]

In principle, the primary line of treatment for haemophilic pseudotumor is to manage acute bleeds with medical treatment, rest, and physical therapy. With advancement, treatment may need to be advanced to low-dose radiation therapy, curettage, or perhaps surgical resection with bone graft.[16]

Complications of compression therapy include skin perforation, abscess and fistula formation, pathologic fractures, compartment syndromes, and joint contractures. In rare situations, rupture of a pseudotumor might result in life-threatening exsanguination. Erosion in an artery can cause severe bleeding. Pseudotumor alter natural anatomy, causing displacements of intestines, ureters, nerves, and blood arteries and this increases the risk of harm during surgery.[17,18]

The most effective treatment for musculoskeletal bleeding in hemophilic patient is prevention with coagulation factor administration from 2 to 18 years of age, preferably at a concentration of less than 1%. Unfortunately, the exorbitant expense of preventive medication prevents 25%-30% of hemophilia patients from receiving it on time. Hemophilia is often regarded as an orphan illness in developed countries due to its low prevalence and expensive treatment costs. This has led to the establishment of interdisciplinary prevention and treatment initiatives.[8]

CONCLUSION

Hemophilic pseudotumor are a common complication of bleeding diseases that doctors should be aware of when treating patients. Early evaluation of compression symptoms including MRI is recommended. Early surgical consultation and factor replacement are crucial if a diagnosis is obtained. In order to give appropriate treatment, it is crucial to identify the potential for missing hemophilic pseudotumor at the time of initial diagnosis.

Declaration by Authors

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