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Case Report

Hemophilic pseudotumor of the ankle: a case report

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Abstract

Hemophilic pseudotumors are little prevalent lesions associated with coagulation factor VIII or IX deficiency that characterizes hemophilia. Their most frequent location are regions such as hips or iliopsoas, whereas distal joints are rare locations. In the present case, we reported the development of a hemophilic pseudotumor related to a conventional anterolateral ankle arthroscopy in a patient with mild hemophilia A who required surgical management for hemophilic arthropathy.

Level of Evidence V; Therapeutic Studies; Expert Opinion.

Keywords: Hemophilia A; Arthrosis; Ankle/abnormalities.

Introduction

Hemophilia A and B are X-linked inherited congenital hemorrhagic disorders characterized by factor VIII and IX deficiency. In Colombia, the 2019 prevalence was 7.5/100 000 inhabitants for hemophilia A and 1.61/100 000 inhabitants for hemophilia B[1].

Patients present with muscular and joint bleeding from an early age, according to disease severity. In some cases, these hemorrhagic events are associated with locally aggressive encapsulated lesions named pseudotumors, whose main control measure is preventing bleeding episodes. The location of pseudotumors is predominantly proximal and axial; thus, it is not often found in acral regions, especially when manifested in a patient with mild deficiency and related to a surgical procedure, like in the case below.

Case report

A 38-year-old patient with history of mild hemophilia A presented with trauma on the right ankle in 2013, subsequent persistent anterolateral pain on the ankle, and feeling of instability. Physical examination revealed pain during tibiotalar mobilization and limited support. Images showed osteoarthritic tibiotalar and subtalar changes and presence of an anterior osteophyte. In view of these findings, the patient underwent arthroscopy for osteophyte resection, synovectomy, and chondroplasty in June 2014, with favorable postoperative outcomes.

In 2018, the patient presented with a progressive enlarging mass at the site of the anterolateral arthroscopy, not related to trauma. Physical examination showed a 3x3cm painless soft mass not adherent to deep planes (Figure 1). Nuclear magnetic resonance imaging (Figure 2) revealed a lesion suggesting hemophilic pseudotumor, which was later resected in 2020 and had its diagnosed confirmed by a subsequent pathological examination (Figure 3).

Pathophysiology

This hematological disorder favors spontaneous bleeding or due to minimum trauma, which is usually is self-limited and is reabsorbed. Of these episodes, 80% occur in the musculoskeletal system[2], and the most frequent are hemarthrosis in the knee, ankle, and elbow, whereas hematomas (10% of the cases) occur in iliopsoas and vastus lateralis muscles[3,4].

After the first episode of hemarthrosis, the events may become persistent or recurrent, which leads to synovial membrane hypertrophy and progressive cartilage injury and is translated into hemophilic arthropathy, manifested in the second and third decade of life[4].

Ankle joint bleeding emerges from the second to the fifth year of life, after children start to walk[5], resulting in a cycle of hemarthrosis and early synovitis, which generates pain and functional limitation associated with angular deformities, especially plantar flexion ones and, later, subtalar joint misalignment and ankle valgus.

Study performed at the Hospital de San José, Fundación de Ciencias de la Salud, Bogotá, Colombia.

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Treatment of hemarthrosis

After the first manifestation of hemarthrosis, the treatment of choice is synoviorthesis at any age[5,6], with an estimated failure rate of up to 25%[7]; moreover, arthroscopic synovectomy is considered a useful alternative to reduce bleeding episodes.

Therefore, treatments of joint disease encompass from minimally invasive joint preservation procedures to cases requiring removal of the joint. Compared with other joints, the ankle presents with a higher incidence of postoperative complications after arthroscopic procedures, among which there are infection and neurovascular lesions.

Pseudotumors

Recurrent bleeding episodes occasionally originate encapsulated hematomas that behave as aggressive lesions causing local destruction, which are named hemophilic pseudotumor or hemophilic cyst[2,3].

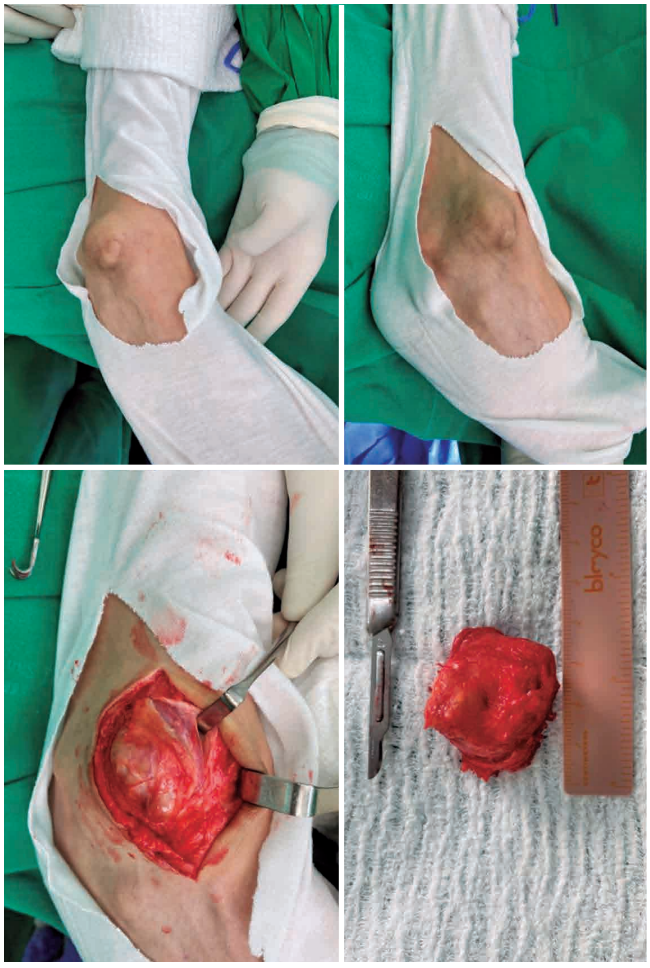

Figure 1. Clinical findings of pseudotumor: 3 x 3 cm mass related to posterolateral arthroscopy.

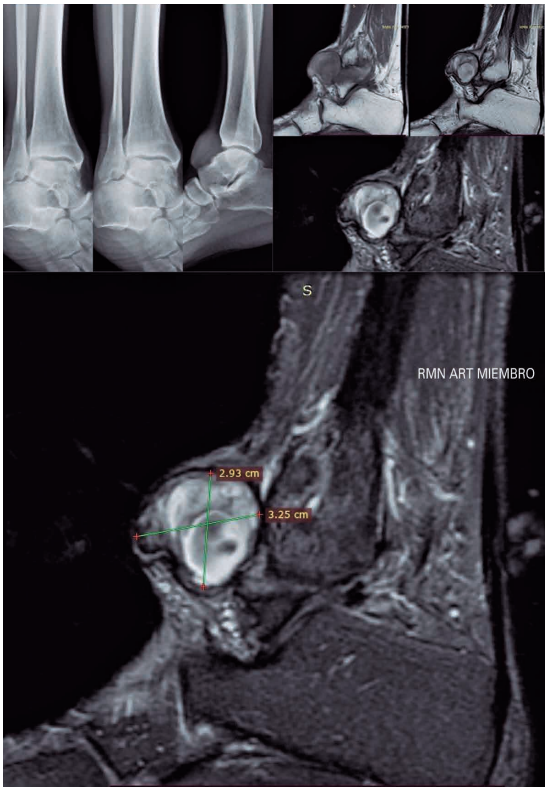

Figure 2. Radiologic images. A) Ankle radiograph showing soft tissue mass in lateral view. A) Nuclear magnetic resonance imaging of the ankle revealing a multilocular, heterogeneous mass.

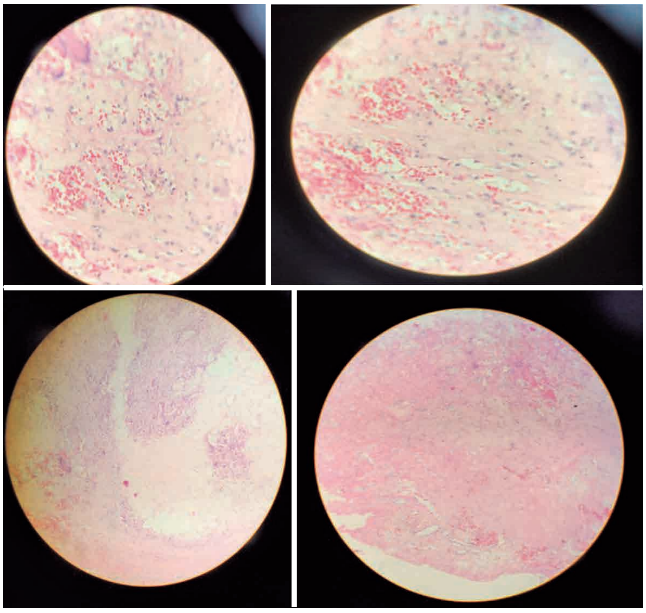

Figure 3. Histopathology of pseudotumor: Red cells with different degrees of lysis, focal hemorrhagic areas, hemosiderin-laden histiocytes, giant cells, and evidence of reaction to foreign body granuloma.

Pseudotumors are manifested as an isolated, unilocular or multilocular lesion[8] that may be secondary to mild, recurrent trauma or trauma, or emerge spontaneously. According to Arnold, they are classified as type A if originated from the soft tissue, type B if originated from the subperiosteal space, and type C when originated from bones. The most frequent location is the femur, followed by pelvis, tibia, and small hand bones, whereas the most frequently affected muscle is the psoas[9].

Pseudotumors have an incidence of 1.14%-2% in patients with severe hemophilia and up to 10% in patients who developed inhibitors[10]; however, no difference was observed in lesion presentation according to coagulation factor deficiency. Most of these lesions are asymptomatic, despite having metabolic activity and progressive growth. Symptoms are derived from compression of underlying structures, bone destruction, and onset of skin fistulas.

Anatomically, pseudotumors are encapsulated hematomas with areas of calcification. Histologically, they contain red cells with different degrees of lysis, subperiosteal bone, and focal hemorrhagic areas with predominantly clotting cells, hemosiderin-laden histiocytes, giant cells, and evidence of reaction to foreign body granuloma. Pseudotumor walls contain a thin inner layer with abundant hemosiderin debris, a thick intermediate layer with fibrous tissue, and an outer layer consisting of muscle fibers, elastin fibers, and blood vessels[2].

Diagnosis is based on clinical findings, whereas imaging assessment determines lesion extent. Computed axial tomography allows for evaluating bone and soft tissue involvement; furthermore, nuclear magnetic resonance imaging shows the specific characteristics of unilocularity or multilocularity and the relationship of the mass with neurovascular structures and joint spaces[6].

Biopsy is not recommended, due to risk of bleeding, in addition to the development of fistula and even of infection[3]. Arteriography may be a diagnostic tool and a therapeutic option if associated with selective embolization.

Treatment of pseudotumors

Currently, there is no consensus on the treatment of pseudotumor; thus, different alternatives are considered, according to patient's findings and resources of each institution. In lesions smaller than 1 cm, intensive replacement therapy has shown good results, and, for larger lesions, associated administration of coagulation factor is considered to reduce lesion size prior to surgical resection.

Radiation therapy provides alternatives in the adjuvant management or definitive treatment, at a total dose of 5-30 Gy, for acral pseudotumors or when surgical resection is contraindicated. Response is observed in the fourth week of treatment, achieving a full cure at 12 weeks. Overall failure rate is 25%, with a lesion recurrence rate of 3.6%, and an increase in size of up to 18%[3].

Although the content of the pseudotumor is mostly solid, the capsule contains the vascular network that is associated with bleeding after its extraction; therefore, embolization of the main vessel for tumors greater than 10cm[3] reduces their size and the risk of bleeding.

Surgical management takes the mass size (<10cm) and consistency into account. In the case of smaller and cystic masses, echography-guided puncture is considered with the benefit of symptom relief, as long as replacement levels are monitored, since this procedure has a recurrence rate of 13%, due to incomplete drainage or bleeding[3].

Solid or mixed masses greater than 10cm with rapid growth and associated with neurovascular involvement, massive and recurrent bleeding, spontaneous perforation, fractures, or failure of symptomatic treatment, require surgical management including excision of the capsule that contains the feeder vessels, except for the cases in which adherence to underlying tissues may lead to greater postoperative bleeding[3]. If bone involvement is greater than 1/3 of the diameter of the segment or bone defect is >5cm, bone graft or structural allograft should be considered.

Complications of pseudotumors

The most frequent complications are recurrence in 15% of the cases and infection in up to 50% of the patients.

Discussion

The presentation of pseudotumors requires a high level of suspicion, since their manifestation in sites such as the pelvis does not allow for a timely detection. The manifestation of the pseudotumor reported herein had early clinical evidence due to scarcity of soft tissue in the perilesional region.

It bears highlighting that the best treatment is preventing musculoskeletal bleeding, based on prophylaxis with administration of coagulation factor, from 2 years to 18 years of age, especially at a concentration lower than 1%[3]. However, 25%-30% of patients with hemophilia do not receive timely prophylactic treatment, due to its high cost. In Colombia, hemophilia is considered an orphan disease, because of its low prevalence and its high treatment cost, which has allowed for the development of multidisciplinary programs of disease prevention and permanent treatment.

Conclusion

Hemophilia is a rare entity, and early diagnosis and prevention of bleeding play a key role in reducing the incidence of hemophilic pseudotumors. These pseudotumors are mostly located in the pelvis and in the mandible and are infrequently found in the ankle, with only one reported case of a pseudotumor in the talus of a girl; thus, the finding of the present case is relevant, especially because it is related to surgical approach.

With regard to the patient reported in the present study, this is a case of mild deficiency that requires replacement of the deficient coagulation factor under demand, which is why episodes of soft tissue hemarthrosis and hemorrhage require timely management, in order to control the bleeding episode and thus resolve the lesion.

The Hospital de San José de Bogotá has a multidisciplinary team for the treatment of primary and secondary manifestations of hemophilia. Orthopedics and hematology services were able to diagnose and treat different musculoskeletal manifestations, such as those described in this report, where we present a case of pseudotumor associated with arthroscopic procedures, considered rare in the reported literature, in addition to a severe manifestation with a challenging medical and surgical treatment.

Authors' contributions: Each author contributed individually and significantly to the development of this article: RRC *(https://orcid.org/0000-002-3817-0609) Conceived and planned the activities that led to the study; CEPL *(https://orcid.org/0000-0001-8197711X) interpreted the results of the study, participated in the review process and approved the final version; ASGF *(https://orcid.org/0000-0003-0296-5263) interpreted the results of the study, participated in the review process and approved the final version; MHST *(https://orcid.org/0000-0001-8752-7080) Conceived and planned the activities that led to the study; CCD *(https://orcid.org/0000-0002-8049-3903) Interpreted the results of the study, participated in the review process and approved the final version; NHA *(https://orcid.org/0000-0003-2120-2181). All authors read and approved the final manuscript. *ORCID (Open Researcher and Contributor ID .

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