Peritesticular scarring: An unusual presentation of hemophilia B

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Abstract

Hemophilia is an inherited clotting disorder that typically presents as spontaneous hemorrhage or prolonged, excessive bleeding following minor trauma. Abnormalities of the genitourinary tract are a rare manifestation of the disease. Here, we report a case of undiagnosed hemophilia B in a teenage boy presenting with worsening testicular pain and a dumbbell-shaped testis. Scrotal ultrasound identified the presence of a hypoechogenic band constricting the left testicle. Surgical exploration of the left testis was performed with release and sampling of the fibrous tunica vaginalis tissue responsible for the testicular deformity. Histopathological analysis revealed evidence of chronic interstitial inflammation with hypocellular keloid-like fiber formation suggestive of old hemorrhage. The procedure was complicated by a delayed scrotal hematoma. Coagulation studies confirmed a prolonged activated partial thromboplastin time and factor IX assay of 5%. Scrotal evacuation with the administration of intraoperative tranexamic acid achieved effective hemostasis. The patient is doing well with ongoing hematology review and prophylactic recombinant factor IX administration.

Keywords: Child, hemophilia, scar, scrotal hematoma, testis

INTRODUCTION

Hemophilia is an X-linked hemorrhagic disorder resulting from deficiencies in or dysfunctional production of clotting factors. Globally, an estimated 350,000 people suffer from some form of the disease. Clinical features range from prolonged bleeding following surgical procedures to spontaneous hemorrhage characteristically involving the joints and muscle tissue.¹⁻³ Genitourinary involvement is rare and typically limited to episodes of benign hematuria.⁴⁻⁵ We describe a case of undiagnosed hemophilia B in a patient presenting with testicular pain and peritesticular scarring. To the best of our knowledge, this is the first reported case of hemophilia presenting as a visceral scar in the form of a testicular band.

CASE REPORT

A 16-year-old boy was referred to pediatric urology with 4 weeks of worsening left testicular pain and the recent development of an overtly dumbbell-shaped left testis. On inquiry, he had presented with ipsilateral groin pain and bruising 3 months earlier, with ultrasound confirming a 15 cm tear of the left iliopsoas muscle. He denied...
any trauma, change in urinary habits, penile discharge, abdominal pain, nausea, vomiting, weight loss, or fevers. He reported no significant personal or family medical history.

General physical examination was unremarkable. Local scrotal examination revealed a centrally pinched, bilobular left testicle [Figure 1]. Palpation was limited by patient discomfort but suggested the presence of normal testicular tissue throughout both poles of the left testis. The lower pole of the testis was tense and tender. There were no palpable testicular masses or groin nodules. The patient was noted to be circumcised, with no other significant findings. Tumor markers (lactate dehydrogenase, human chorionic gonadotropin, and alpha-fetoprotein) were normal. Ultrasound investigation confirmed the presence of a faintly hypoechoic band constricting the left testis. The left lower testicular pole was noted to be moderately hyperechoic when measured against the upper pole, but still well vascularized. A 2 mm hyperechoic lesion was also identified in the left lower pole [Figure 2]. Testicular sonography and Doppler flow studies revealed normal intratesticular flow and the absence of a classic whirlpool sign suggestive of testicular torsion.

Scrotal exploration with testicular release and Jaboulay procedure was performed. A fibrous band in the left tunica vaginalis was identified intraoperatively, divided, and sent for histopathological assessment [Figure 3]. No other macroscopic testicular abnormalities were identified. Samples of the left tunica vaginalis fluid and left testicular appendix were also collected for analysis. Cytology reported normal tunica vaginalis fluid with the presence of benign mesothelial cells, macrophages, and lymphocytes. No malignant cells were present. Microscopic analysis of the excised band showed areas of hypocellular thick keloid-like fiber formation, with adjacent chronic interstitial inflammation and hemosiderin deposits, suggestive of previous trauma and hemorrhage [Figures 4 and 5]. The left testicular appendix was histologically normal.

On postoperative day 2, the patient developed a large left hemiscrotum. Clotting investigations reported an international normalized ratio of 1.2, prothrombin time at the upper limit of normal at 16s, a prolonged activated partial thromboplastin time of 45s, and fibrinogen of 4.0 g/L. He was returned to theatre for scrotal hematoma evacuation and exploration, requiring tranexamic acid and 3 units of fresh frozen plasma intraoperatively to control bleeding. Subsequent special coagulation orders revealed a factor IX assay of 5%, diagnostic of mild-to-moderate hemophilia B. Scrotal swelling was markedly improved postoperatively, with only mild bruising at the base of the penis and scrotum. The patient was referred to the pediatric hematology department for ongoing management of his factor IX deficiency. With prophylactic recombinant factor IX treatment, he has made a satisfactory recovery and has had no further bleeding episodes.
The involvement of the genitourinary tract is uncommon in hemophilia. It typically involves spontaneous or provoked hemorrhage resulting in hematuria, renal colic, or postprocedure bleeding and is rarely the presenting symptom of the underlying coagulation disorder.\textsuperscript{[4,5]} To the best of our knowledge, this is the first reported case of hemophilia presenting as a visceral scar in the form of a testicular band.

Hoffman \textit{et al.} utilized a mouse model to demonstrate that cutaneous wound healing is impaired in hemophilia B.\textsuperscript{[6]} A few clinical studies and case reports have also identified a likely predisposition to keloid scarring in patients with factor XIII deficiency.\textsuperscript{[7,8]} There are currently no reported cases of visceral scar associated with hemophilia.

In our hemophilia B patient, we hypothesize that scrotal microtrauma caused hemorrhage and aberrant inflammatory responses resulting in the deposition of keloid-like scar tissue. This resulted in testicular pain and development of a bilobular left testicle, a unique, genitourinary presentation of disordered coagulation.

For young male patients presenting with testicular pain and unexplained, unusual physical findings, bleeding diathesis is a rare but important differential diagnosis.

\textbf{Declaration of patient consent}

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient’s legal guardian has given the consent for images and other clinical information to be reported in the journal. The patient’s legal guardian understands that name and initials will not be published, and due efforts will be made to conceal identity, but anonymity cannot be guaranteed.

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\textbf{Conflicts of interest}

There are no conflicts of interest.

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