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Acute scrotum caused by an extraspermatic venous thrombosis associated with G20210A prothrombin gene mutation in a pediatric patient: A case report

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ABSTRACT

Thrombosis of an extraspermatic blood vessel has been reported to be an unusual cause of acute scrotum. We describe the case of a 7-year-old boy who presented with an acute painful left scrotal swelling. Imaging demonstrated a heterogeneous left scrotal mass, which was surgically resected. The anatomopathologic study revealed a thrombosed vascular structure. Three months later, another extraspermatic venous thrombosis occurred and was also resected. A complete thrombophilia study was done. A G20210A prothrombin gene heterozygous mutation was identified as the cause of the thrombotic event. A venous thrombosis should be considered as a possible cause of acute scrotum in European Caucasian patients, due to its increased prevalence. We reviewed the G20210A prothrombin gene mutation, which is associated with thrombosis.

Key Words: Extraspermatic; thrombosis; G20210A gene; thrombophilia.

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Introduction

The G20210A mutation in the 3′ UTR region of the prothrombin gene causes high blood levels of prothrombin, which manifest as venous thrombosis. The risk of venous thromboembolism in carriers of this mutation is increased two- to fourfold [1]. This mutation is rare, but its prevalence is higher in the Caucasian population, especially in Southern European countries.

In children, the G20210A prothrombin gene mutation most frequently manifests as an arterial thrombosis, especially in the central nervous system. There have been around 18 case reports of acute scrotum caused by spermatic venous thrombosis [2,3]. We have previously observed one case of scrotal vein thrombosis, but it was not associated with this mutation. However, since the G20210A mutation is responsible for thrombus formation in unusual locations, it must be considered a potential cause of thrombotic acute scrotum.

Case report

A 7-year-old Caucasian boy without relevant past medical history presented to the pediatric urology outpatient clinic with a history of acute painful left scrotal swelling, treated with ibuprofen without relief. A purple mass was found in the left hemiscrotum (Fig. 1). It was studied with ultrasonography and magnetic resonance imaging (MRI). AFP and BCG values were normal, but LDH was high (561 U/L). Ultrasonography revealed an extrascrotal cystic complex lesion and a probably thrombosed varicocele (Fig. 2). MRI demonstrated a 9-mm left scrotal mass, probably attached to the epididymis. The findings were not suggestive of malignancy.

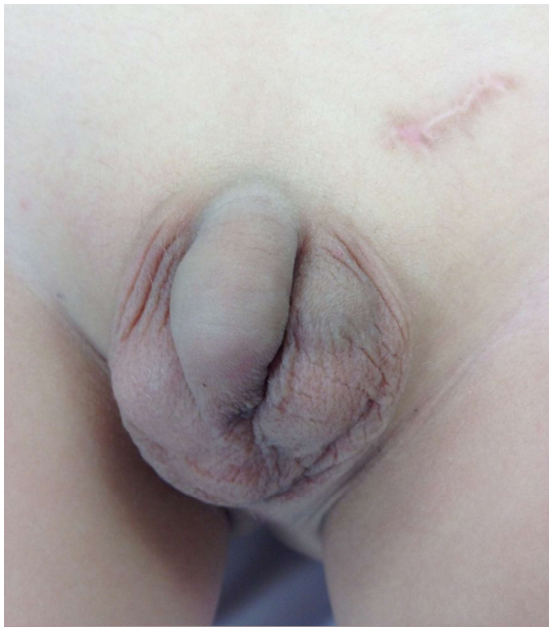

Fig. 1. A purple mass was evident at both the first thrombotic event and the recurrent episode.

Surgical resection of this mass was carried out. The anatomopathologic study revealed scrotal adipose tissue with vascular congestion and fibrin deposits, compatible with venous thrombosis (Fig. 3). At postsurgical control after 3 months, repeat ultrasonography evidenced a nodular structure of 9x8 mm in the upper pole of the left testicle.

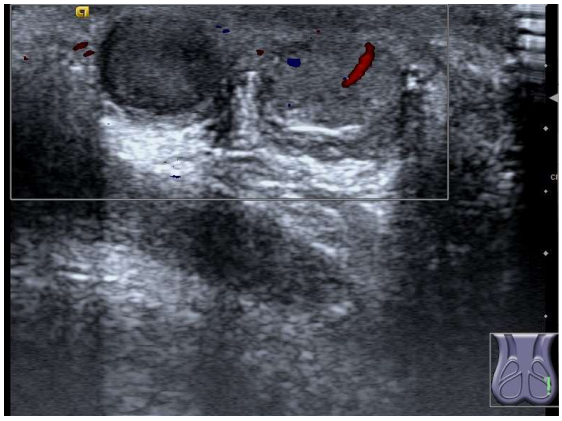

Fig. 2. Left testicle, with normal vascularization, adjacent to a hypoechoic nodule measuring 9x8 mm, without color Doppler signal, that is compatible with a thrombosed vein.

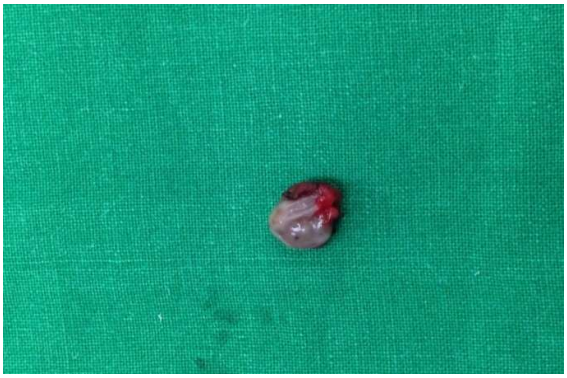

Fig. 3. The left scrotal resected mass, which was found to be a venous thrombosis.

This mass was also surgically resected and was identified as another venous extraspermatic thrombosis. As the patient presented venous thrombosis at an unusual location, a complete thrombophilia study was done. This study showed the patient to be a carrier of a G20210A heterozygous mutation of the prothrombin gene. Every other laboratory test was normal. The hematology service concluded that in this situation it was not necessary to chronically anticoagulate the

patient. They advised antithrombotic prophylaxis only in high-risk situations such as surgery or prolonged hospitalization. The patient remained stable at 9 months following the last surgical resection.

Discussion

The G20210A mutation consists of a single-nucleotide polymorphism (substitution of adenine for guanine) at position 20210 in the 3′-UTR non-coding region of the prothrombin gene on chromosome 11. As this mutation is located in a non-coding region, it does not affect prothrombin structure or function [1]. G20210A prothrombin gene mutation causes higher prothrombin messenger RNA and protein expression [2]. Excessive prothrombin blood levels result in hypercoagulability and an elevated risk of venous thromboembolic events.

The prevalence of the G20210A prothrombin gene mutation has been found to be 3–17% in European Caucasians with venous thromboembolic events (VTE) and 1–8% in healthy controls. This prevalence is highest in Southern European countries. The mutation is very rare in Asians, Africans, native Americans and Australians. A high prevalence has been found in Hispanic populations outside of Europe owing to their European ancestry [3].

The G20210A prothrombin gene mutation has been associated with early-onset stroke [4,5], deep venous thrombosis [6], myocardial infarction [7] and recurrent spontaneous abortion. In children younger than 2 years of age, this mutation is more often associated with arterial thrombosis. On the other hand, children older than 2 years are at higher risk of VTE [8].

Including the current case, a thrombotic event has been reported to be the cause of acute scrotum in 19 cases. In all of these cases, thrombosis of a varicocele or the spermatic vein was observed. G20210A prothrombin gene mutation was reported in none of them. Factor V Leiden mutation has been associated with this pathology [9]. The clinical manifestations consist of acute testicular pain and swelling. Most cases occur on the left side [10]. The presence of a purple mass in the posterior aspect of the left hemiscrotum oriented our diagnostic approach and was the reason for performance of ultrasonography and MRI. These imaging studies were decisive in establishing the diagnosis. In the context of a testicular mass of unknown etiology, we decided to conduct a surgical exploration. Recurrence of the thrombotic event led us to carry out a thrombophilia study, which also proved decisive in identifying the etiology of the pathology in this young patient.

In most cases of this nature, conservative management is indicated. Anticoagulation is suggested as prophylaxis for surgery, and prolonged bed rest [10].

Conclusion

Acute scrotum has a wide spectrum of etiologies. Extraspermatic vein thrombosis is a rare cause of acute scrotum. It should be considered in European Caucasian patients, especially those from Southern European countries, due to the increased prevalence of the G20210A prothrombin gene mutation. Further studies are needed to standardize the management of these patients.

Compliance with ethical statements

Conflicts of Interest: None.

Financial disclosure: None.

Consent: All photos were taken with parental consent.

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