A case of hereditary angioedema who presented with difficulty in urination and globe

Summary
Hereditary angioedema (HEA) is a disease characterized by decreased levels or function of C1 esterase inhibitor (C1-INH). The symptoms of HEA in pediatric age group generally consist of recurrent episodes of soft tissue swelling. These symptoms can be transient, subtle, and varied in severity. Genitourinary system is rarely affected in this disease. Here, a three-year-old girl who presented with angioedema on her hands, fingers, and face, and difficulty in urination and globe is reported. The aim of this case is to focus on this rare disease, hereditary angioedema, which presented with difficulty in urination and urinary globe.

Case report
One day, a three-year-old girl with HEA was admitted to our emergency pediatric department with swelling of right forearm and urinary globe, and difficulty in urination (figure 1). She didn't have dysuria, fever or pelvic pain. Urinary globe, mild external genital swelling and right forearm angioedema were examined in her physical examination. Her urine analyses, and induce increased permeability. C1-INH is a serine protease inhibitor (serpin), also known as SERPING1, that blocks the activity of some complement components (e.g. C1r, C1s, Mannose binding lectin-associated serine protease; MASP-1 and MASP-2). C1-INH also controls contact-kinins, coagulation, and fibrinolytic cascades (6). In this paper, we report a case with rarely manifested hereditary angioedema who presented with difficulty in urination and globe.

Introduction
C1-INH deficiency can be genetic or acquired. HAE is a rare, life-threatening condition manifested by acute attacks of facial, laryngeal, peripheral or genital edema. The estimated prevalence of HAE is between 1 in 10,000 and 1 in 150,000 people (1,2). The genetic HEA deficiency is due to mutations in one of the two alleles of the C1-INH gene that result in reduced protein levels in plasma (type I HEA) or in normal protein levels but always in reduced function (type II HEA). A third type has also been reported, occurring exclusively in women who have normal C1-INH levels and function (Type III HEA). It is estimated that 20% to 25% of HAE cases are caused by spontaneous mutations in patients with no family history of the disease (3,5). The development of angioedema in C1-INH-deficient patients involves the inappropriate generation of kinins (particularly bradykinin) that stimulate vascular smooth-muscle relaxation and induce increased permeability.
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Our patient was admitted with forearm swelling, difficulty in urination and globe, without hematuria and dysuria, and her symptoms were decreased with the administration of C1 esterase inhibitor concentrate.

Her laboratory tests (urine, blood) were normal for the other etiologies of urinary globe and difficulty in urination. In summary, we have described a case of type I HEA with most likely de novo mutation. The patient had recurrent episodes of angioedema, urinary globe and difficulty in urination, which resembled manifestations of urinary infections and other etiologies of urinary globe. HAE should be taken into consideration for the differential diagnosis of urinary globe etiology.

References


