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SHORT COMMUNICATION

Peripheral Angioedema and Upper Airway Edema in Young Woman

Gargantilla Madera P*, García Tobaruela A and Belda Bilbao L

Department of Internal Medicine, University Francisco of Vitoria, Spain

***Corresponding author:** *Pedro Gargantilla, Department of Internal Medicine, Hospital of El Escorial Madrid, University Francisco of Vitoria, Spain, E-mail: pgargantillam@gmail.com*

Keywords

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C1 inhibitor (C1-INH) deficiency is a rare syndrome clinically characterized by recurrent localized and self-limiting acute edema episodes in different locations [1] or angioedema without urticaria. It can involve the skin, upper respiratory airways and abdomen.

There are two main types: Hereditary (C1-INH-HAE) and acquired (C1-INH-AAE). We present the clinical characteristics of a patient with angioedema secondary to C1-INH-AAE type II.

A 38-year-old woman, without family history of angioedema, suffered several self-limiting acute edema episodes (localized in upper/lower limbs). One month after, she presented an upper airway edema associated with glottis edema, which was treated in emergency department with corticosteroids, without improvement.

Routine blood tests performed were normal, but the immunological study reveled low C4 concentrations (2.6 mg/dL), normal antigenic C1-INH (14.2 mg/dL), low C1-INH function (25.7%), normal serum C1q (157.95 µug/dL) and presence of anti-C1-INH IgG autoantibodies. Sequencing of the C1-INH and F12 genes revealed no pathogenic variants. She was treated with 1000 units of C1 inhibitor plasma concentrate (Bertinert, CSL-Behring) with a good response and no adverse events.

We report the case of a patient who developed C1-INH-AAE. Our initial diagnosis was C1-INH-HAE, since C1q was normal, a mutation in the C1-INH gene is not found in 5-10% of families with C1-INH-HAE and up to 25% of patients can have a spontaneous mutation [2]. But positive anti-C1-INH antibodies enabled us to make a more accurate diagnosis of C1-INH-AAE type II.

This disease was first described by Caldwell, et al. in 1972 and it is due to deficiency of C1 esterase inhibitor, hyperactivation of the classical pathway of human complement and angioedema symptoms mediated by bradykinin released by inappropriate activation of the contact-kinin system [3].

The annual incidence is 1.09 individuals per 100,000 inhabitants in Spain and the onset of C1-INH-AAE is most common after the fourth decade of life. Persons of any race can be affected by C1-INH-AAE. Men and women are equally affected.

C1-INH-AAE, much less prevalent than C1-INH-HAE, and is often associated with lymphoproliferative disorders (type I) and/or the presence of anti-C1-INH autoantibodies (type II) [1].

C1-INH-AAE is first suspected in patients aged 40 or above who present with recurrent cutaneous and/or mucosal angioedema without urticaria, without an evident triggering factor, and without family history of angioedema.

The edema produced in C1-INH-AAE has been linked to the release of kinins, their activation leads to vascular leakage, which accounts for the typical non-puritic, non-urticarious, and non-erythematous edema of C1-INH-AAE in contrast to the traditional histamine-associated itchy wheal and flare presentation of allergic edema.

The clinical presentation mostly involves peripheral angioedema of the head, neck and extremities, fol-

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lowed by abdominal pain. The diagnosis is based on clinical features and characteristic alterations of laboratory tests [4]. Medications that can increase the frequency and/or severity of attacks in AAE include the following: Estrogen-containing medications (hormone replacement therapy and contraceptives), tamoxifen and angiotensin-converting enzyme inhibitors.

The average time to diagnosis from the onset of symptoms has been reported between 2.3 and 2.48 years [2,3]. In our patient the diagnosis was realized in 5 months. In this moment our patient is asymptomatic, and she have not complications.

The differential diagnosis should include intestinal occlusion syndrome, hereditary angioedema and histamine-induced angioedema (of allergenic or nonallergenic origin) generally associated with urticaria [2].

The prognosis for patients with AAE is variable, and, in most cases, it depends on control of the underlying disorder. Angioedema-related fatalities derive from laryngeal edema. Compared with the general population, patients with acquired angioedema have a higher incidence of B-cell malignancies [2]. In the first patients reported by Caldwell AAE occurred in presence of lymphoma. Monoclonal gammopathy of uncertain significance have been reported with high frequency in association with AAE.

Assessment and protection of the upper airway is the first and most important management issue in the patient with an acute attack involving any part of the airway. Based on the efficacy of replacement therapy with plasma-derived C1-INH in reverting laryngeal edema.

A small percentage of patients with C1-INH AAE become less responsive to C1-INHRP over time, requiring higher doses to control symptoms. In these patients we must to use the newer therapies for bradykinin-mediated angioedema (ecallantide and icatibant) [5]. Rituximab can be an alternative treatment in cases of C1-INH-AAE associated with C1-INH autoantibodies.

Angioedema prophylaxis is performed using antifibrinolytic agents and attenuated androgens (stanozolol, danazol) with antifibrinolytic agents providing somewhat better results [2].

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