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Case Report

Hereditary Angioedema: An Unusual Cause of Abdominal Pain

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Abstract

Introduction: Hereditary angioedema (HAE) is a genetic disease that affects about one in 50,000 people. It's characterized by recurrent episodes of edema and associated with a deficiency of C1-inhibitor.

Case Report: We report a very rare case of hereditary angioedema, presenting as recurrent abdominal pain. A 37-year-old patient consults for recurrent abdominal pain, chronic vomiting and edema of the face and hands. Those symptoms started in childhood and disappear in a few days. Abdominal ultrasounds and CT scans are normal. All upper endoscopies performed were normal. The C4 complement level and the concentration of C1 esterase inhibitor were low. HAE type I was retained and the patient started tranexamic acid substitution with a resolution of symptoms.

Conclusion: The clinician should think about hereditary angioedema when other causes of recurrent abdominal pain are ruled out, especially in patients with a history of recurrent episodes of edema and proceed to the investigation of C3 and C4 levels then of C1 esterase inhibitor if levels of C4 are low.

Keywords: *Hereditary Angioedema; Abdominal Pain; C4 Complement; Prophylaxis*

Abbreviations

HAE: Hereditary Angioedema; AE: Angioedema; CT scan: Computed Tomography Scan; C1-INH: C1-Inhibitor; Complement C3: Complement Component 3; Complement C4: Complement Component 4

Introduction

Von Quicke was the first who described the angioedema in 1882, and Sir William Osler had created the term hereditary angioneurotic edema in 1888 [1].

HAE is an autosomal dominant genetic disorder characterized by a mutation in the C1-INH gene. Patients affected with HAE have recurrent episodes of swelling of the skin and mucous membranes which evolve in a few hours and persist for a few days [2].

Case Report

A 37-year-old patient, presented for recurrent atypical abdominal pain, chronic vomiting and episodes of edema of the face and hands. Those episodes started in childhood and disappear spontaneously in a few days without general physical health deterioration. He had two brothers with the same symptoms.

The CT abdominal scans were normal as well as many upper endoscopies. The C4 complement level and the concentration of C1 esterase inhibitor measured by a two functional test were low (less than 10%).

HAE type I was retained and the patient started tranexamic acid substitution with a resolution of symptoms.

Discussion

Hereditary angioedema is a very rare disease. It's estimated to affect 1 in 10 000 to 1 in 50 000 IN in the general population, and there is no evidence of sex, ethnicity in the prevalence [1].

HEA is a genetic disorder characterized by mutations of the SERPING1 gene, responsible for increased activation of C4 levels with an excess generation of bradykinin levels. The activation of the Hageman factor (factor XIIa) leads to the cleavage of prekallikrein, which active kallikrein-kinin cascade and form kallikrein, which release bradykinin peptide [3]. Bradykinin is an inflammatory mediator that causes clinical manifestations (Figure 1) [4].

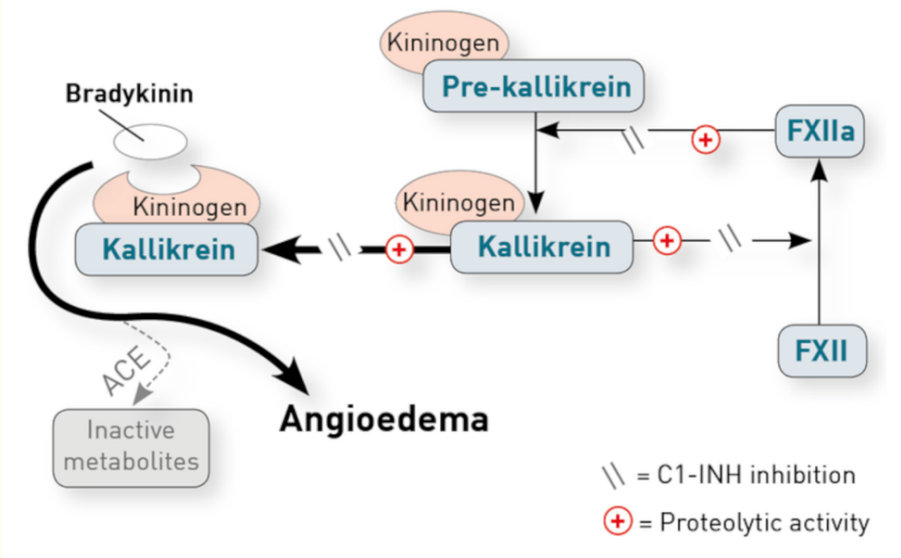

Figure 1: *Physiopathology of bradykinin-induced angioedema [3].*

Symptoms generally begin in childhood and increases after puberty. These patients suffer from recurrent severe swelling of the skin and mucous membranes [5]. It is estimated that one in two individuals will have an upper airway attack in their life It is estimated that one in two individuals will have an upper airway attack in their life [6].

The diagnosis of patients with a clinical suspicion of HAE; the measures of C4, C3 and C1-INH levels are necessary, ideally during an episode, and the measures should be repeated by a second analysis within 1-3 months [7].

The treatment of HAE consists of prophylaxis of acute attacks in the short-term and reduces the frequency and severity of the angioedema crisis in the long-term [2].

The prophylaxis of acute attacks in the long-them is required in patients with a high risk of developing laryngeal edema, or with one crisis monthly. It relies on the use of antifibrinolytics agent, or synthetic 17- α-alkylated androgens. Short-term prophylaxis consists of the administration of C1-INH concentrate 1-1,5 hours before the surgery or any invasive procedure. In the countries where C1-INH concentrate is not available, a high dose of androgen should be administrated for 5 - 7 days [8].

The management of acute attacks consists of the administration of C1-INH concentrate (Berinert, 20U/kg intravenous), or Icatibant injection (Firazyr, 30 mg subcutaneous) while contacting the local national referent [9]. If a C1-INH concentrate or Icatibant are not available, attacks should be treated with plasma kallikrein inhibitor (Kalbitor) or with fresh frozen plasma [2].

Conclusion

The clinician should think about hereditary angioedema when other causes of recurrent abdominal pain are ruled out, especially in patients with a history of recurrent episodes of edema and proceed to the investigation of C3 and C4 levels then of C1 esterase inhibitor if levels of C4 are low. There are national associations for HAE patients and their families around the world to facilitate the management of HAE.

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