Case report

Hereditary Angioedema: A Case Report

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Abstract

Hereditary angioedema (HAE) is a extremely rare disease, which is caused by deficiency or dysfunction of C1-esterase inhibitor. A 28 year old woman presented to emergency department with edema at face, eyes and around the mouth and tongue. Antihistaminic and corticosteroid were given again after monitorization of the patient. Fresh frozen plasma was given at a dose of 10 ml/kg. She was discharged after resolution of complaints during the follow-up period.

Key word: *Hereditary angioedema, C1-esterase inhibitor, edema*

Introduction

Hereditary angioedema (HAE) is a extremely rare disease with autosomal dominant inheritance, which results from deficiency or dysfunction of C1-esterase inhibitor produced by the liver and involved in the regulation of kinin-kallikrein, complement and fibrinolytic system. Moreover, there is an acquired type of HAE caused by C1 esterase inhibitor antibody [1-3]. It is equally seen in females and males. Prevalence of this disease is estimated as 1:10.000-50.000 [2]. HAE is accounted from 2% of all angioedema cases [3]. The disease is characterized by recurrent subcutaneous and submucosal edema at any localization of the body such as respiratory system, skin or gastrointestinal system.

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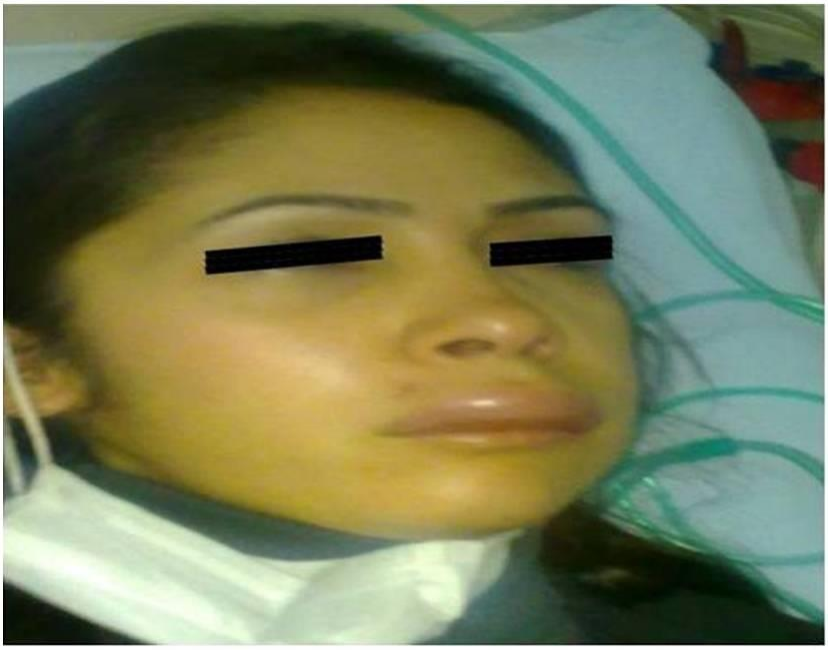
Case Report

A 28 year old woman presented to emergency department with edema at face, eyes and around the mouth and tongue. In the physical examination: blood pressure; 110/60 mmHg, heart rate; 119 beat/min, body temperature; 36.2 ^{0}C, respiratory rate; 24/min. Diffuse edema developed at face, eyes, lip and tongue mucosa (figure-1).

There was no edema at the larynx and vocal cords. She had normal respiratory findings, whereas there was no significant finding other than tachycardia in cardiac examination. Antihistaminic and corticosteroid were given again after monitorization. She was given fluids for hydration. No factor such as food or insect bite was found in relation to etiology. It was found out that the patient's complaints

increases at winter and with stress. Family history indicated similar complaints are present in her mother and aunt. In history, she stated that she had 1 to 3 attack per year ; C1 esterase inhibitor levels were found to be low during detailed tests in other facilities, when hereditary angioedema diagnosis was established. Fresh frozen plasma was given at a dose of 10 ml/kg. The patient was admitted to emergency department for a 24 hour follow-up. She was discharged after resolution of complaints during the follow-up period

Figure-1. Diffuse edema in face, eyes, lip and tongue mucosa



Discussion

Hereditary angioedema, a result of C1 esterase inhibitor deficiency, has a clinical course characterized by recurrent, non-pitting edema without itching and urticaria. C1 esterase inhibitor levels is an important regulatory protein for classical complement pathway, fibrinolytic pathway, and kinin-kallikrein system. Deficiency of this protein causes increased vascular permeability and edema through release of bradikinin-like mediators due to increased kallikrein [4].

Of the patients with HAE, 75% have family history as in our case [5]. It has

been reported that angioedema attack might be triggered by factors including disorders causing tissue injury, infection, tooth extraction, surgical interventions such as tonsillectomy, trauma, menstruation, stress, oral contraceptive use, anxiety and fatigue [6]. Frequency of attack varies between individuals from weekly to yearly intervals [7]. In our patient, it was found that attacks initiated at adolescent period and she had 1 to 3 attacks per year.

Routine laboratory evaluations are normal in HAE. In our case, they also were normal in accordance to literature. Screening test is C1 esterase inhibitor levels concentration for HAE. Definitive diagnosis was made by measuring C1 esterase inhibitor levels. Enzyme should be assessed in quantitative and functional manner [1, 7-9].

Subcutaneous edema and abdominal attacks, which are most commonly seen symptoms in HAE, are not life threatening. Although larynx edema is rarely seen, it may cause death through causing sudden airway obstruction [3]. In HAE, follow-up is sufficient in mild cutaneous and abdominal symptoms, as they may spontaneously resolve. If there is a larynx edema or severe abdominal attack, it should be treated. The optimal and most rapid approach is to use intravenous C1 esterase inhibitor concentrates. Symptoms relieve 30-60 minutes after injection. However, it is difficult to provide this preparation in the first attack. Thus, the optimal alternative is fresh frozen plasma in patients presented with first attack. It is used at a dose of 10 ml/kg [10]. We administered fresh frozen plasma to patient because of C1 esterase inhibitor concentrates were not available.

In conclusion, it should be kept in mind that HAE may be present in patients frequently presenting to the emergency department with diffuse, recurrent edema and anaphylaxis-like presentation, and if they also have family history; thus, such patients should be evaluated for HAE.

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