Case Report

# Navigating the swells: A case report of hereditary angioedema

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## Abstract

Hereditary angioedema (HAE) is a rare genetic disorder characterized by recurrent episodes of localized edema caused by a deficiency or dysfunction of C1 inhibitor (C1-INH). This case report presents the clinical features, diagnostic evaluation, and management of a 23-year-old man with HAE. We discuss the challenges of diagnosing and treating this condition, emphasizing the importance of early recognition and appropriate therapeutic interventions.

**Keywords:** C1 inhibitor deficiency, hereditary angioedema, recurrent angioedema

## Introduction

Hereditary angioedema (HAE) is a rare, autosomal dominant genetic disorder characterized by recurrent and unpredictable episodes of localized edema in various body parts.[1] It is caused by a deficiency or dysfunction of C1 inhibitor (C1-INH), a regulatory protein involved in controlling the complement and coagulation systems.[2] The condition leads to uncontrolled activation of bradykinin, resulting in increased vascular permeability and angioedema formation.[3] HAE affects approximately 1 in 50,000 individuals worldwide[4] and can manifest at any age, with onset typically occurring during childhood or adolescence.[5]

The clinical manifestations of HAE are diverse and often overlap with other conditions, making diagnosis challenging. Despite significant advancements in the understanding and management of HAE, the condition remains underdiagnosed and undertreated, leading to potential complications and decreased quality of life for affected individuals. Early diagnosis and appropriate management are crucial to mitigate the impact of HAE on patients and prevent life-threatening complications.

## Case Presentation

The patient is 23-year-old man with a medical history significant for recurrent episodes of angioedema affecting various sites, including the face, hands, and abdomen. The patient reported a positive family history of similar symptoms in his mother. He first experienced episodes of angioedema at the age of 12, which initially presented as swelling of the lips and tongue following a respiratory tract infection. Over the years, the episodes increased in frequency and intensity. The patient also reported that the episodes were usually triggered by minor trauma, emotional stress, or infections.

In the current episode, he presented to us with facial swelling predominantly lip swelling [Figure 1]. The episode was associated with nausea and diffused dull aching abdominal pain. There was no history of cough, dyspnoea, or fever. On examination during

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**Figure 1:** Photograph of the patient with hereditary angioedema

this episode, the patient displayed facial swelling, which included the lips and mild periorbital region swelling [Figure 1]. The abdominal examination revealed diffuse tenderness with minimal guarding. There was no evidence of urticarial rash or erythema.

Laboratory investigations revealed low levels of C1-INH (quantitative assay by the radial immunodiffusion method), that is, 173 mg/L. The diagnosis was further supported by low C4 complement levels, that is, 0.9 gm/L. Based on the clinical presentation and laboratory findings, the patient was diagnosed with HAE type 1.

He was advised for intravenous C1-INH concentrate therapy; however, due to financial restrain, the patient could not afford the therapy. So, he underwent administration of fresh frozen plasma (FFP), which contains C1-INH. FFP can be used as an acute treatment option in patients without access to specific C1-INH concentrate. However, FFP administration is associated with a risk of adverse reactions and may not be as effective as C1-INH concentrate. He was also given tranexamic acid, an antifibrinolytic agent, which acts by preventing the breakdown of blood clots and reducing bradykinin release. For symptomatic relief, he had an anti-histaminic, a short course of steroids, anti-emetics, and analgesics.

He was counselled to identify and avoid triggers, such as trauma, stress, and infections, which could potentially precipitate angioedema attacks. He was instructed to seek immediate medical attention during the acute attack and asked for regular follow-up appointments. Additionally, the patient's family members were offered genetic testing and counselling to identify potential carriers of the HAE-causing mutation.

## Discussion

HAE is a rare and potentially life-threatening disorder, making its diagnosis and management challenging. HAE can be broadly classified into three types:[6]

1. Type 1 HAE: Characterized by a quantitative deficiency of C1-INH protein.
2. Type 2 HAE: Characterized by a dysfunctional C1-INH protein.
3. Type 3 HAE: A less understood form associated with normal C1-INH levels, but with mutations in the coagulation factor XII gene (FXII-HAE).

Clinical manifestations of HAE include recurrent episodes of angioedema affecting various sites, such as the skin, gastrointestinal tract, and upper airways.[7] Skin-related angioedema commonly involves the face, lips, tongue, and extremities, while gastrointestinal symptoms can lead to severe abdominal pain, nausea, and vomiting. Laryngeal edema is a potentially life-threatening complication that may lead to airway obstruction.[7]

### Diagnosis

1. Clinical presentation: HAE is characterized by recurrent and localized episodes of edema affecting various body parts. Symptoms often include facial swelling, abdominal pain, and potential airway obstruction.
2. Family history: A positive family history of HAE is common.
3. Laboratory tests: Low C1-INH levels and decreased C4 complement levels support the diagnosis.[8,9]

### Management[10]

1. Acute attacks: Acute HAE attacks are treated with C1-INH concentrate, which helps control the symptoms by inhibiting bradykinin release. Bradykinin receptor antagonists (icatibant, ecallantide) can also be used as alternative treatments.
2. Prophylactic therapy: Long-term prophylaxis is recommended for patients experiencing frequent or severe attacks. Attenuated androgens[9] (danazol, stanozolol) or antifibrinolytic agents can be used for prophylactic management.
3. Trigger avoidance: Identifying and avoiding triggers, such as trauma, stress, and infections, may help reduce the frequency of HAE attacks.
4. Genetic counselling: Patients and their families should consider genetic counselling to understand the inheritance pattern and potential risks for future generations.
5. Antifibrinolytic agents: Tranexamic acid acts by preventing the breakdown of blood clots and reducing bradykinin release.
6. FFP: It contains C1-INH and can be used as an acute treatment option in patients without access to specific C1-INH concentrate. However, FFP administration is associated with a risk of adverse reactions and may not be as effective as C1-INH concentrate.
7. Symptomatic management: Anti-histaminics and corticosteroids can be used as adjunctive therapy to manage the symptoms of HAE. However, these agents are not primary treatments for HAE attacks.

Management strategies should be individualized, and the risks and benefits of each treatment should be carefully considered

in the context of the patient's medical history and overall health. Regular follow-up appointments are crucial to monitor treatment efficacy and adjust the management plan as needed.

## Conclusion

HAE is a rare genetic disorder characterized by recurrent episodes of localized edema. Timely diagnosis, differentiation from other forms of angioedema, and appropriate management are essential to minimize the burden of the disease. Clinicians should be familiar with the diagnosis, available laboratory tests, and treatment options to provide optimal care for patients with HAE.

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### Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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### Conflicts of interest

There are no conflicts of interest.

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