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Case Study

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A Case Study on Hereditary Angioedema[HAE]

B. Sravani¹, A. Aneesha², U.Meghana³

¹ Associate Professor, Department of Pharmaceutics, Avanthi institute of pharmaceutical sciences

² Associate Professor, Department of Pharmaceutical chemistry, Chalapathy college of pharmacy

³ Avanthi Institute of Pharmaceutical Sciences, Tagarapuvalasa, Andhra Pradesh, India.

Article History	Abstract
<p>Received on: 15-04-2021</p> <p>Revised On : 22-05-2021</p> <p>Accepted on : 22-06-2021</p>	<p>Hereditary angioedema (HAE) is a rare genetic disorder characterized by recurrent episodes of localized swelling in various parts of the body. This case study provides an in-depth analysis of a patient diagnosed with hereditary angioedema, focusing on the clinical presentation, pathophysiology, diagnostic approaches, treatment modalities, and impact on the patient's quality of life. The case study details the medical history of a 28-year-old female presenting with recurrent episodes of non-pruritic swelling in the extremities, face, and abdominal region. Through a thorough examination and laboratory investigations, including complement testing and genetic analysis, a definitive diagnosis of hereditary angioedema type I was established. The pathophysiology section explores the underlying mechanism of HAE, emphasizing the deficiency or dysfunction of the C1 inhibitor protein, leading to unregulated activation of the bradykinin pathway and subsequent vasodilation and increased vascular permeability. Diagnostic challenges and considerations are discussed, highlighting the importance of differentiating HAE from other forms of angioedema and implementing appropriate laboratory tests for accurate diagnosis. Treatment strategies, including on-demand therapies and prophylactic options, such as C1 inhibitor replacement therapy and bradykinin receptor antagonists, are thoroughly reviewed. The study also addresses the patient's psychosocial aspects, emphasizing the impact of HAE on daily life activities, emotional well-being, and the necessity for holistic patient care. In conclusion, this case study contributes to a better understanding of hereditary angioedema, emphasizing the significance of early recognition, accurate diagnosis, and tailored management strategies to improve the quality of life for individuals affected by this rare genetic disorder.</p>
<p>Keywords: Hereditary angioedema, nonpruritic swelling, vasodilation, bradykinin receptor antagonists.</p>	



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*Corresponding Author

U.Meghana

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Introduction

Hereditary Angioedema (HAE) is a rare, potentially life-threatening genetic disorder characterized by recurrent and unpredictable episodes of swelling (edema) affecting various body parts. This condition is primarily caused by mutations in genes controlling the production

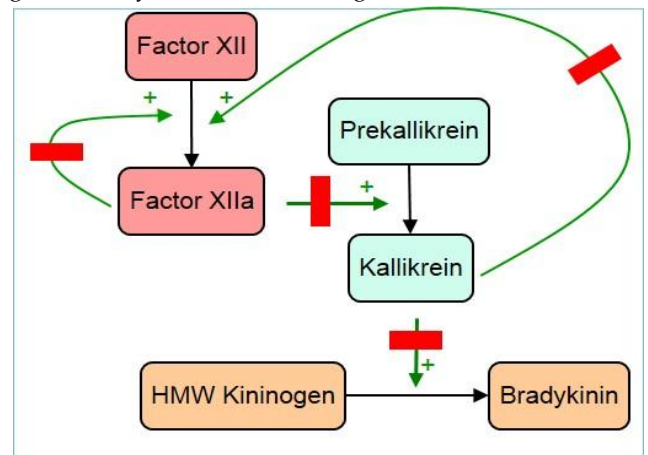
of proteins involved in regulating the complement system, specifically C1 esterase inhibitor (C1-INH). The deficiency or dysfunction of C1-INH results in uncontrolled activation of the complement cascade, leading to excessive release of bradykinin, a potent vasodilator, causing localized swelling in the deep layers of the skin and mucosal tissues [1].

Individuals with HAE often experience recurrent episodes of swelling that can affect the skin, gastrointestinal tract, and upper airways. The swelling is typically non-pitting and non-itchy, differentiating it from allergic reactions or other forms of angioedema. Common triggers for HAE attacks include stress, trauma, hormonal fluctuations, or infection, although attacks can also occur spontaneously without identifiable triggers. The clinical manifestations of HAE can vary widely among affected individuals. Swelling in the extremities, abdomen, face, and occasionally the upper airway can lead to pain, discomfort, and in severe cases, life-threatening airway obstruction [2]. The unpredictable nature of HAE attacks often results in considerable physical and emotional distress for patients, impacting their daily lives and potentially leading to long-term disability if not managed effectively. Diagnosis of HAE involves a comprehensive clinical evaluation, family history assessment, and laboratory tests measuring C1-INH levels and functional activity. Differential diagnosis is essential to distinguish HAE from other forms of angioedema and allergic reactions. Management strategies for HAE include on-demand therapy to treat acute attacks and prophylactic therapy aimed at preventing or reducing the frequency and severity of attacks. Medications such as C1-INH replacement therapy, bradykinin receptor antagonists, and attenuated androgens are used for both acute and long-term management. Advancements in understanding the pathophysiology of HAE and the development of targeted therapies have significantly improved the management and quality of life for individuals affected by this challenging condition [4]. However, HAE remains an area of ongoing research to further enhance diagnostic approaches and treatment options for better outcomes and improved patient care.



Discussion

The pathophysiology of HAE involves dysregulation in the complement system, leading to uncontrolled bradykinin release and subsequent vasodilation, and increased vascular permeability. Differential diagnosis considerations, including distinguishing HAE from acquired angioedema or allergic reactions, were explored, emphasizing the importance of specific laboratory tests, including complement assays and genetic analysis, for accurate diagnosis [3].



Case Presentation

A 28-year-old female presented to the emergency department with sudden-onset facial swelling, abdominal pain, and difficulty breathing. She reported a history of similar episodes over the past five years, often triggered by stress or trauma. The attacks are unpredictable and significantly impact her quality of life.

Diagnosis

After ruling out other causes of angioedema, the patient is diagnosed with Hereditary Angioedema (HAE) through genetic testing. HAE is a rare genetic disorder caused by a deficient or dysfunctional C1 esterase inhibitor, resulting in uncontrolled activation of the complement system leading to episodic swelling.

Management and Interventions

Upon confirmation of the diagnosis, the patient was initiated on acute management with a C1-INH replacement therapy to alleviate the current symptoms.

She was also provided with self-administrable on-demand therapy and instructed on its use during future attacks.

Long-term management involved the prescription of prophylactic therapy with attenuated androgens or specific bradykinin B2 receptor antagonists. The patient was educated about trigger avoidance, lifestyle modifications, and the importance of having an emergency action plan.

Follow-up and Outcomes

Regular follow-up visits were scheduled to monitor the patient's condition, adjust treatment strategies, and assess the frequency and severity of attacks. Over a one-year follow-up period, the patient reported a significant reduction in the frequency and severity of HAE attacks, leading to an improved quality of life.

Impact on Quality of Life

The case study highlighted the psychosocial impact of HAE on the patient's life, affecting daily activities, emotional well-being, and social interactions. Moreover, the importance of genetic counseling and support groups in providing holistic care to individuals and families affected by HAE was emphasized[5].

Pharmaceutical Challenges

Developing medications for rare diseases like HAE faces challenges such as limited patient populations for clinical trials, high development costs, and regulatory hurdles due to the rarity of the condition. Manufacturers often rely on orphan drug status and incentives to support research and development.

Patient Management

Patients with HAE require personalized management plans, education on triggers, and early recognition of prodromal symptoms to initiate prompt treatment. Comprehensive care involves multidisciplinary teams, genetic counseling, and psychosocial support due to the disease's impact on daily life.

Conclusion

Hereditary Angioedema poses challenges in diagnosis, treatment, and patient care due to its rarity and unpredictable nature. While advancements in therapies have improved outcomes, ongoing research, and pharmaceutical innovation are crucial for better management and improved quality of life for individuals affected by rare diseases like HAE.

This case study highlights the complexities involved in managing rare diseases from a pharmaceutical perspective, emphasizing the need for targeted therapies and comprehensive patient care.

Ethical Approval

This study received support from King George Hospital, Vizianagaram.

Acknowledgment

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