# The burden of hereditary angioedema

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**Hereditary angioedema (HAE)** is a rare disorder, affecting **1 in 50 000 individuals**. The disease is genetic and caused by a deficient or dysfunctional protein (the C1-Inhibitor) which is involved in controlling multiple biological processes in the body. The true prevalence could be much higher, due to the rarity of the disease and associated misdiagnoses.

HAE is characterized by episodic and unpredictable swelling (HAE attacks) in the hands, feet, face, gastrointestinal tract and airways, among other locations. HAE attacks can be highly **debilitating**, painful, and potentially life-threatening, and can disrupt patients' and carers' daily lives, leading to long-term anxiety and depression.[1,2]

## HAE symptoms and triggers

- **7 years** is the median time to diagnosis.
- While there is no clear cause of attacks; physical activity, hormonal changes, viral infections, trauma and stress can be **triggers.**[4]

**Most people living with HAE experience their first attack before age**

**18**[3]

**50%**

people living with HAE will experience at least one potentially fatal airway attack during their lifetime.[5]

## The long journey to diagnosis

Despite advancements diagnosis delays persist, especially when there is no family history: in approximately 25% of cases, HAE is the result of spontaneous mutations[7]. HAE is often misdiagnosed as **allergies**[6]. Misdiagnoses can lead to unnecessary surgeries.

**Quality of life impact**

This journey can be frustrating and overwhelming for people living with HAE, demanding time and determination to obtain an accurate diagnosis. While a family history of HAE can support faster diagnosis, the emotional burden of the disease remains. Negative treatment experiences of relatives and potential deaths in the family can even add to the burden.

**Increasing awareness of this rare disease and educating healthcare professionals while investing in centres of excellence can help fast-track diagnosis, referrals, and personalized care aligned with global guidelines.**[8]

## Treatment optimisation

HAE care is a **lifelong journey**, often requiring months to personalize treatment. Constant adaptations are needed to accommodate for changing **hormone** levels, aging, and **comorbidities** and treatment approaches may be varied depending on life stages. In **women**, menstruation and pregnancy can make HAE symptoms worse.

**Quality of life impact**

The need for constant treatment vigilance places a psychological strain on people living with HAE, while carers also endure emotional strain from living in a state of constant alertness.

**Governments must collaborate with all stakeholders to ensure that every patient has access to therapies to be used across the three treatment strategies: on-demand treatment (to manage HAE attacks as they occur), short-term prophylaxis (to reduce the risk of attacks in trigger situations), and long-term prophylaxis (to prevent attacks).**

## Living with HAE

Modern preventive treatments significantly reduce attack frequency and swelling severity, offering patients a chance at a more **normal life.** However, no treatment fully eliminates attacks[9], and some may be poorly tolerated or require frequent administration.

**Quality of life impact**

Patients may tolerate occasional mild attacks as a **trade-off** for stability, often preferring familiar treatment over uncertainty. For those attack-free for a while, a breakthrough attack can be a distressing reminder of **past struggles.**

**Enabling shared and informed decision-making is key to ensure treatment regimens reflect patients' evolving lifestyles and needs.**

## Spotlight: accessing modern long-term prophylaxis

Long-term prophylaxis (LTP) has been transforming patients' lifes by helping to prevent attacks. While there is no cure for HAE yet, **treatments that may extend attack-free periods** can improve quality of life and address HAE's unpredictable nature. However, these may remain out-of-reach due to eligibility thresholds or unequal access.

**Incorporating the perspectives of people living with HAE in decision-making is essential for identifying ongoing unmet needs and recognizing the benefits of expanding access to new treatment options, ultimately enhancing the quality of life for people living with HAE.**

Living with HAE is a lifelong journey, but personalized treatment and innovative solutions bring hope, helping patients manage their disease and regain confidence to live their lives without limitations.

Collaboration among all stakeholders is essential to address the hidden HAE disease burden and unlock access to innovation that improves patient quality of life.

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