Taking a Closer Look

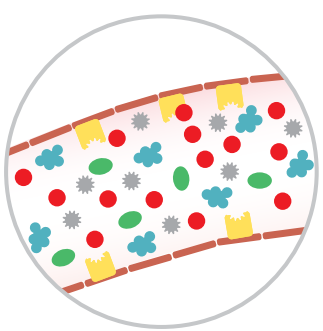
at Hereditary Angioedema (HAE)

HAE: a rare, genetic condition

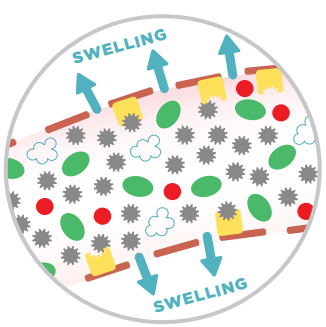
HAE is a rare, debilitating, and potentially life-threatening disease that affects approximately **1 in 50,000 people**. HAE swelling attacks are the result of a complex cascade that occurs in the body involving certain proteins, such as C1 esterase inhibitor, plasma kallikrein, and bradykinin.

Most people with HAE don't have enough of a protein called C1 esterase inhibitor, or this protein does not work as it should. When there is not enough functioning C1 esterase inhibitor, the activity of plasma kallikrein increases. Too much plasma kallikrein activity triggers the release of bradykinin. Increased bradykinin levels cause blood vessels to release fluid and results in the localized swelling and pain of an HAE attack.

A person without HAE



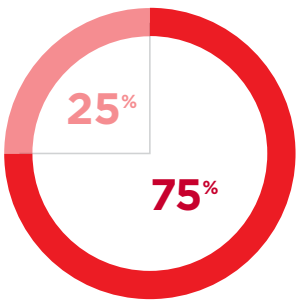
A person with HAE during an attack (untreated)



C1 esterase inhibitor
Missing/dysfunctional C1 esterase inhibitor
Bradykinin
Kallikrein
Blood vessel wall
Fluid
Red blood cell
Bradykinin B2 receptor

Understanding the causes of HAE

HAE is called "hereditary" because it can be passed down through families.

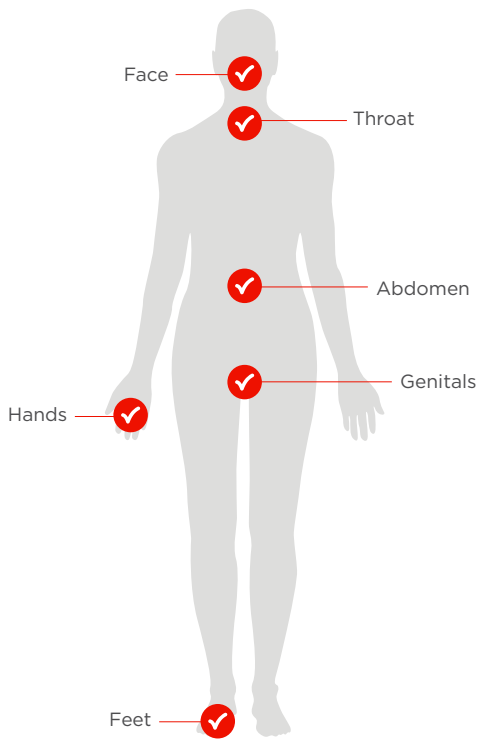


- Percentage of people with HAE who inherit it
- Percentage of people with HAE who have a spontaneous genetic mutation

50% If one parent has HAE and the other does not, each child will have a **50% risk** of inheriting the condition.

HAE is unpredictable

HAE can be challenging to manage because symptoms may change over time. Because of this, people with HAE don't know when their next attack will come. Attacks of swelling can be painful, vary in frequency and severity, and can happen in different parts of the body. The most common places include:



An HAE attack can start in one location then spread to another before resolving.

HAE attacks can be debilitating and potentially life-threatening

HAE attacks can affect different parts of the body in different ways. And the next attack may be nothing like the last.

Throat, or larynx

- In a 2006 study of 201 people with HAE, 54% had experienced a throat attack
- A throat attack, otherwise known as a laryngeal attack, causes swelling in the throat, which can interfere with breathing and create a potentially life-threatening situation
- May cause other symptoms, such as voice changes and difficulty swallowing

Stomach, or abdomen

- Abdominal attacks can cause severe pain and may be accompanied by vomiting and/or diarrhea
- Some people with HAE who had untreated abdominal attacks had to stay in bed between 24 and 50 hours*

*In a survey of 23 people with HAE.

Skin, or subcutaneous tissue

- Swelling of the skin usually affects the hands, face, feet, or genitals
- May be accompanied by redness, but the area usually doesn't itch

HAE attacks can happen at any time without warning

Before an HAE attack, some people experience early warning signs, also called prodromal symptoms, such as a tingling sensation, rash, fatigue, or nausea. Although HAE attacks often happen without a known trigger, they can sometimes be caused by:

- Minor trauma (an injury or a shock to the body)
- Stress
- Dental procedures
- Infection
- Surgery
- Hormonal changes

Characteristics of HAE attacks

- An untreated attack usually lasts 3 to 5 days
- Some attacks affect just one part of the body, while others can affect multiple parts
- On average, a person with HAE who is not on preventive treatment will experience 2 to 4 attacks per month

Additional questions about HAE

Does HAE affect men and women?

Men and women are both affected, but women tend to present with symptoms more frequently than men do. This might be because hormones may influence HAE attacks. For children, symptoms may become more frequent and/or severe during puberty.

What kind of testing is available for HAE?

Laboratory tests are available to confirm a diagnosis of HAE. Doctors may perform blood tests to measure specific proteins (C4 and C1 esterase inhibitor) that may be lower than normal or dysfunctional in people with HAE.

How is HAE treated?

Treatment for HAE can include:

- Preventive therapy (also known as prophylactic) that involves taking medicine routinely to help prevent or reduce the frequency and severity of HAE attacks
- On-demand treatment (also known as acute treatment) used to treat symptoms of an HAE attack

The 2020 US Hereditary Angioedema Association (HAEA) guidelines recommend that people with HAE should have at least 2 doses of an on-demand treatment with them at all times and discuss preventive treatment at every doctor visit.

What organizations can I get involved with to learn more about HAE?

There are many ways you can learn more about HAE and help raise disease awareness. You can join the HAEA, a nonprofit organization that serves and supports people with HAE. Visit **HAEA.org** to learn more.

HAE can impact your life in many ways

If you're living with HAE, you're probably aware of the many ways attacks and their unpredictability can limit your life. You're not alone. Here's what people with HAE are saying about the impact of their symptoms:

In a 2017 survey of 445 people with HAE...

50% said they experienced anxiety

In a 2007-2008 survey of 457 people with HAE...

69% reported that symptoms prevented them from applying to certain types of jobs

58% reported that HAE impacted their career advancement

Answer and discuss

Ask yourself these questions, and talk about them with your doctor:

- How many times in the past year have you had to cancel social plans because of an HAE attack or fear of a future attack?
- What effects do your HAE attacks have on your job or your career?

Be prepared for an HAE attack

Fill out your HAE attack plan with your doctor to share it with those in your life who may be unfamiliar with the condition. This tool contains helpful information about HAE as well as your treatment plan, which can be used in the event of an emergency or unexpected attack.

In addition to the attack plan, it can help to carry a medical information card in case of an emergency.

Fill out the card provided below and keep it on hand.

Medical Information Card

I have hereditary angioedema (HAE), a rare and potentially life-threatening genetic disorder. Please present this card to a medical professional on my behalf.

Name: _____

Address: _____

Home Phone: _____

Cell: _____

Talking to your doctor about HAE

When discussing treatment options with your doctor, be sure to share details about your attack frequency and severity, as well as how HAE impacts your life. Recording this kind of information will help you prepare for the conversation.

Emergency Contacts

Name: _____

Relation: _____

Phone: _____

Name: _____

Relation: _____

Phone: _____