



## Hereditary Angioedema (HAE)

Hereditary angioedema (HAE) is a rare inherited disorder which is characterized by painful swelling that can appear in various parts of the body. This disease is also sometimes referred to as C1 Inhibitor Deficiency. It most commonly affects the hands, feet, face, abdomen, urogenital tract, and upper respiratory tract. The inflammation can be disfiguring, debilitating, or in the case of laryngeal attack, life-threatening. If left untreated, HAE can result in a mortality rate as high as 40%, primarily due to upper airway obstruction or suffocation in undiagnosed patients.

HAE is estimated to occur in at least 6,000 people in the US. It is a rare inherited disease that can span multiple generations. Because it is a genetic disease, children of HAE patients have a 50% chance of inheriting the disease.

The underlying cause of HAE is decreased production, or production of a non-functional form of a protein found in your body called C1 inhibitor (C1-INH), which is involved in mediation of the inflammatory response. There are three systems (Contact, Complement, and Fibrinolytic) that exist in your body. Normal levels of C1 inhibitor serve to regulate these systems to modulate inflammation and ensure HAE attacks do not occur. However, HAE patients cannot produce enough functional C1 inhibitor; this deficiency allows these systems to go unchecked, resulting in the episodes of swelling that are classified as HAE attacks.

HAE has been reported in all races and ages, with equal occurrence amongst males and females. A patient's quality of life and ability to carry out normal daily activities are often significantly affected as a result of experiencing HAE attacks. Most HAE attacks last 2 to 5 days, resulting in 20 to 100 days of inactivity per year. Due to this inactivity, HAE patients frequently find their ability to accomplish daily activities significantly reduced.

HAE attacks account for approximately 15,000 to 30,000 emergency department (ED) visits per year. And, because symptoms of HAE can mimic a surgical emergency, about one-third of patients with undiagnosed HAE have been reported to have undergone unnecessary surgery during abdominal attacks, because the symptoms very closely resemble appendicitis and other medical conditions that frequently require surgery.

HAE attacks occur without warning, and are sometimes times brought on by a common trigger. In women, the number and severity of attacks may increase during menstrual periods especially during puberty. Common triggers include:

- Trauma
- Surgical Procedures
- Autoactivation
- Dental Procedures
- Emotional Stress
- Menstruation
- Use of birth control pills
- Infections

Unfortunately, HAE is frequently misdiagnosed, with some patients reporting that they were misdiagnosed for periods of 20 years or longer. In the U.S., approximately 68% of patients known to suffer from HAE were initially misdiagnosed. Since the symptoms of HAE attacks closely resemble symptoms of other disorders, patients are commonly misdiagnosed with allergies,

appendicitis, stress disorders, adverse reactions to insect bites, stomach ulcers and other ailments before receiving an accurate diagnosis of HAE.

For more information on HAE, visit the U.S. HAE Association's website at: [www.haea.org](http://www.haea.org)