Hereditary Angioedema (HAE)

What? Who? When? Where? Why? How?

What is HAE?

- HAE is a rare genetic disorder characterized by recurring painful attacks of swelling in different areas of the body. The severity of the attacks can vary between patients and throughout an individual patient's lifetime. Attacks can also be disfiguring and when they occur in the throat, can be life-threatening due to the risk of suffocation.[1]

Who gets HAE?

- HAE Affects between 1 in 10,000 to 1 in 50,000 people,[2, 3]
- Most people with HAE have inherited it from a parent. About 20-25% of cases of HAE occur spontaneously, without previous family history.[4]
- Children of someone with HAE have a 50% chance of inheriting HAE.[5]
- HAE affects men and women[6], with higher rates in women, and people of all ethnic groups[7] at approximately the same rates.

When does HAE occur?

- Inherited HAE symptoms can start at different ages, even in children just a few years old. Half are symptomatic by age 7. For some, symptoms only appear in their 30's or 40's.[8]
- Majority of HAE swelling attacks are unpredictable.[9] Some can have attacks as frequently as every three days while others have attacks less often.[10]
- Swelling attacks generally occur without warning, though in some cases an attack is preceded by symptoms such as fatigue, irritability, or a pink rash.[11]
- Attacks usually worsen steadily over the first 24 hours then gradually subside over the next two to three days.[12]
- Some attacks can be as short as four hours and others can last as long as five days.[13]
- In general, people with HAE are incapacitated by attacks from 20 to 100 days per year.[14]
- Certain triggers or events can bring on the attacks such as an injury, surgery, dental procedures, anesthesia, stress, infection, fluctuating hormone levels, menstruation or use of oral contraceptives.[15]

Where in the body does HAE affect?

- Almost any part of the body can be affected by a swelling attack, but attacks most commonly occur in the face, extremities (arms, hands, legs, feet), genitals, abdomen, or throat. [16]
- Attacks can be localized in one area of the body or can spread to another location before resolving.[17]
- Attacks in the abdomen and intestines are usually the most painful and can cause severe vomiting, diarrhea and cramping that can be mistaken for other conditions, such as appendicitis, that require surgical intervention.[18]
- Attacks leading to swelling in the throat are life threatening medical emergencies that require prompt attention due to the risk of suffocation.[19]

Why does HAE happen?

- HAE is the result of a problem with a protein called C1 esterase inhibitor, or abbreviated as C1-INH.[20]
- In Type I HAE, accounting for 85% of cases, C1-INH level is decreased or not present at all.[21]
- In Type II HAE, accounting for 15% of cases, C1-INH is present but does not function properly.[22]
- A rare form, HAE with Normal CI-INH level and function has been described.[23]
- Problems with C1-INH means that the body's systems that control inflammation are no longer regulated, eventually opening gaps in blood vessel walls through which fluid leaks to tissues causing swelling.[24]

How is HAE diagnosed, prevented and treated?

- Because HAE and acute abdominal conditions are rare and the symptoms can be similar to other medical conditions, including allergic reactions, diagnosis is often difficult and patients may be incorrectly treated.[25]
- Laboratory analysis of blood samples or genetic testing is required to establish the HAE diagnosis. There are two specific blood tests that confirm HAE: C1 inhibitor quantitative (antigenic), which measures the amount, and C1-inhibitor functional, which measures how it works. [26]
- Four Health Canada approved treatments are available for HAE in Canada.[27]
- There are treatments which exist to help reduce symptoms of an attack but need to be taken as early as possible to be most effective. This is best accomplished when patients are trained and equipped to self-administer by intravenous infusion at home or by subcutaneous injection as soon as an attack starts.[28]
- Treatments can also be administered between attacks to help prevent them.[29]
- New Canadian guidelines for the medical management of HAE were published in October 2014 and are available online.[30]

For More information visit:

www.HAECanada.org/about-hae/resources-links

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