DISCOVER HAE

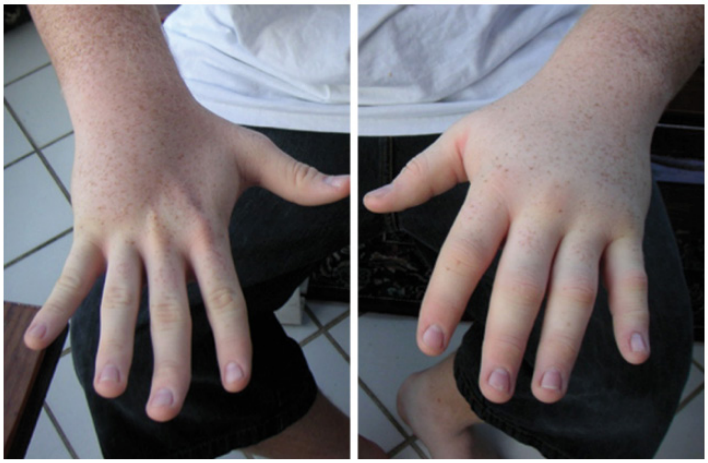
Genetics of Hereditary Angioedema

Understanding the Family Aspect of Hereditary Angioedema (HAE)

HAE: A family affair

HAE is a rare genetic disorder that affects about 1 in 10,000 to 1 in 50,000 people in the world. People with HAE have a defect in the gene that makes C1 esterase inhibitor (C1-INH), an important protein in the blood that helps prevent swelling.

HAND SWELLING DURING AN HAE ATTACK



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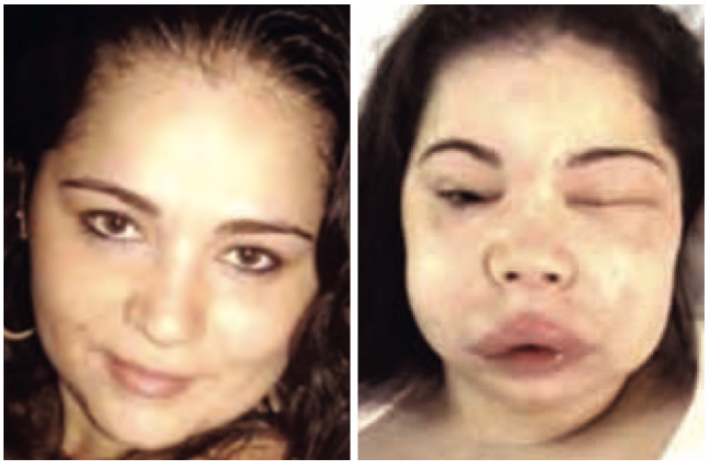
FACIAL SWELLING DURING AN HAE ATTACK



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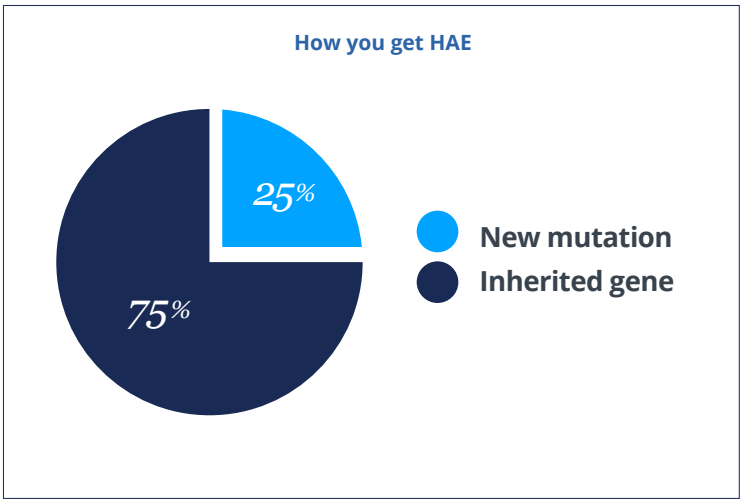
GENETICS CAN PLAY A BIG PART IN HAE

HAE is called hereditary because it can be passed on from a parent. Seventy-five percent of people with HAE get it through the family connection. However, 25% have no family history. These people develop HAE from a spontaneous genetic mutation that causes the disease. Once a person has HAE, there is a chance that it can be passed down to future generations.



Even when people from the same family have HAE, they can each experience the disease in different ways. For example, one person with HAE might experience much more severe symptoms than another family member who also has HAE. The symptoms also vary with time for one patient.

MISDIAGNOSIS IS COMMON

Because HAE is a rare disease and its symptoms often mimic those of other diseases, people with HAE are commonly misdiagnosed. A 2010 survey of 313 patients showed that up to 65% of patients with HAE may have been misdiagnosed with another condition prior to receiving their HAE diagnosis.

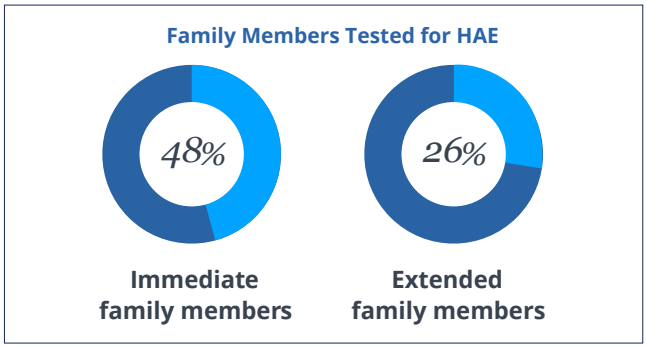
HAE attacks can be mistaken for allergic reactions or appendicitis. Some people have even been referred for psychological evaluation.

When the same survey asked 125 patients living in the US about previous surgeries, 19% underwent an unnecessary surgical procedure, including removal of the appendix.

If you have a family history of HAE, it's important to give this medical information to your doctor. Laboratory findings are a critical part of the diagnostic process and may rule out other types of angioedema. Your doctor's knowledge of your family history can actually improve your likelihood of receiving an earlier and accurate diagnosis.

SCREENING IS IMPORTANT

In the survey of 313 patients with HAE, respondents reported an average of 2 immediate and 2 extended family members who were diagnosed with HAE. However, even though HAE is a hereditary disease, family members often go untested. Respondents in the same survey noted that only 48% of their immediate family members and 26% of their extended family members had been tested for HAE.



In fact, a thorough knowledge of your family's health history can not only help cut down on unnecessary procedures, but also help ensure proper education and management of HAE. This includes identification and avoidance of triggers, preparation for acute attacks and ways to properly react to life-threatening complications, such as airway obstruction.

HAE IN FUTURE GENERATIONS

People with HAE may have questions about how the disease is inherited. HAE is considered an autosomal dominant disease, which means an individual has a 50% chance of inheriting the disorder if one of his or her parents has it. Most people with HAE have their first attack in childhood or adolescence.

The decision to be tested for the disorder is different for every family. Talk to your doctor. Together, you can decide what the best options are for your family.

Diagnosing HAE

CLINICAL DIAGNOSIS OF HAE

As part of determining whether you have HAE, your doctor will conduct a physical examination and ask you questions about your swelling attacks. He or she will look for the following signs:

- Recurrent swelling that doesn't itch or look like hives
- Other family members who have been diagnosed with HAE, or show the symptoms (this is the case for 75% of the people who are diagnosed with HAE)
- Symptoms that start in childhood/adolescence
- Recurrent attacks of pain in the abdomen
- Occurrence of swelling in the airway

- Swelling that doesn't get better after taking antihistamines, glucocorticoids, and/or epinephrine
- Signs or symptoms in the affected area before an attack occurs

Your doctor may also ask about your family health history, which is especially important if you have a family history of HAE.

TYPES OF HAE

There are 3 types of HAE. People with Type I have low levels of C1-INH. About 85% of HAE cases are Type 1.

People with Type II have normal or even elevated levels of C1-INH that do not function properly. About 15% have Type II.

HAE with normal C1-INH (previously known as Type III HAE) is extremely rare. People with this type of HAE have normal levels of C1-INH that works the way it should. Scientists are not certain what causes HAE attacks in this type.

LABORATORY TESTS FOR DETECTING HAE

Most cases of angioedema are not HAE. Often, allergic reactions or another medical condition can cause swelling attacks. Doctors require laboratory tests or genetic testing of blood samples to establish the HAE diagnosis.

The following tests can help confirm an HAE diagnosis:

- C4 level
- C1-INH level, antigenic
- C1-INH, functional

People with HAE Types I and II have low levels of C4, so many doctors use the C4 test as a first screening step. If a person has low levels of C4, a doctor may decide to do additional C1-INH tests to further confirm the HAE diagnosis and determine the type of HAE.

Diagnosing HAE with normal C1-INH can be more complicated. Instead of the blood tests used to confirm Types I and II, a doctor will make a diagnosis on other findings, like symptoms, family history, how you respond to certain medications and genetic testing.

THE ROAD AFTER DIAGNOSIS

Although a confirmed diagnosis of HAE may come as a relief to people who have spent years experiencing attacks without knowing the cause, it will also bring a whole new set of questions and expectations. For instance, you may ask: What exactly is HAE? Will my disease worsen as I get older? What therapies are available for people with HAE? Your doctor can answer all of your questions and discuss therapy options with you and your family. Be honest with your doctor about your symptoms and your experiences with any types of medications. Maintaining an open relationship with your healthcare provider is an important aspect of managing your HAE.

TO LEARN MORE, TALK TO YOUR DOCTOR OR VISIT WWW.DISCOVERHAE.COM.