Hereditary Angioedema
Frequently Asked Questions

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For patient or carer support contact HAE Australasia.

Q 1: What is hereditary angioedema?

Hereditary angioedema, also called HAE, is a very rare and potentially life-threatening genetic disorder that occurs in around one in 50,000 people.

'Hereditary' means that this condition runs in families because it is a genetic disorder. If one parent has HAE, their children have a 50% possibility of inheriting this disease.

Less commonly, there may be no family history of the condition, as HAE may occur "spontaneously". Around 20% of cases result from people who have a spontaneous mutation of the particular gene at conception. Subsequently, these people can pass the defective gene to their children.

'Angioedema' is a term used to describe swelling of the tissues. There are many other causes of this type of swelling, the most common being allergic causes.

People with HAE have unpredictable, recurrent and rapid swellings (HAE attacks) throughout their life. HAE attacks affect daily life and can be life threatening.

HAE has been classified as a primary immunodeficiency (PID). Inborn errors of immunity (IEI) include PID, and are a group of more than 550 potentially serious chronic medical conditions.However, unlike other PID/IEI, people with HAE do not have an increased risk of infections.

Q 2: What are the symptoms of HAE?

Symptoms of HAE attacks include episodes of:

- Angioedema (swelling) in various body parts including the hands, feet, face and airway. These episodes can last for days causing great discomfort and interfering with daily life.
- Excruciating abdominal pain, nausea and vomiting that is caused by swelling in the gut wall.
- Airway swelling which can be dangerous as it may lead to death by closing over the airway.

The age when swellings begin in people with HAE varies. In one study, half of the patients reported onset of symptoms by seven years of age and over two-thirds became symptomatic by thirteen years of age. There is an increased frequency of attacks during puberty or adolescence.

If you are diagnosed with HAE, remember to have other family members tested, as HAE runs in families.

Q 3: What causes HAE?

People with HAE have a defect in the gene that controls a blood protein called C1-Inhibitor:

- The role of C1-Inhibitor protein helps to regulate the complex biochemical interactions of blood systems involved in immunity, inflammation and blood clotting.
- The genetic defect in people with HAE results in production of either an abnormal or non-functioning C1-Inhibitor protein.
- Abnormal or absent C1-Inhibitor protein does not perform its important regulatory function, which causes a biochemical imbalance. This produces other chemicals that can cause tiny blood vessels to leak fluid into

surrounding tissue, causing swelling or oedema.

Q 4: How is HAE diagnosed?

Diagnosis of HAE is usually considered due to typical swellings or a family history suggestive of the condition.

There are three blood tests that are used to confirm HAE - a screening test called C4 and specific tests of the level and function of the C1-inhibitor protein.

There are three recognised forms of HAE:

- **HAE Type I** is the most common form which affects around 85% of people with HAE - blood tests show low quantitative levels of C1-inhibitor protein.
- **HAE Type II** affects the other 15% of people with HAE who have normal or elevated levels of C1-inhibitor protein, but the protein does not function properly.
- **HAE Type III** is an extremely rare form of HAE where the levels of C1-inhibitor protein are normal – there are different genetic defects causing this form.

Q 5: How is HAE treated?

HAE cannot currently be cured, although scientists are working on gene therapy which may result in exciting possibilities for the future.

Currently there are modern treatments available that can be used to:

- Treat an acute HAE attack, to reduce severity and duration of the attack.
- Prevent HAE attacks in the first place.

Some dental and surgical procedures can be dangerous for people with HAE, so it is important that protective treatment is used beforehand to limit the risk of HAE attacks.

The treatment options should be discussed with your clinical immunology/allergy specialist to enable you to have the most appropriate treatment for your circumstances.

It is important to note that acute HAE attacks do not respond to antihistamines, corticosteroids or adrenaline.

Q 6: Where can I find support and further information?

For more detailed information on HAE and for patient or carer support, visit the HAE Australasia website: haeaustralasia.org.au

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For more information go to www.allergy.org.au/patients/immunodeficiencies

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