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Hereditary angioedema

Hereditary angioedema is a rare but serious problem with the immune system. The problem is passed down through families. It causes swelling, particularly of the face and airways, and intestines (causing severe abdominal pain).

Causes

Angioedema is swelling that is similar to hives, but the swelling is under the skin instead of on the surface. Angioedema typically is not itchy while hives are very itchy.

Hereditary angioedema (HAE) is caused by a low level or improper function of a protein called the C1 inhibitor. It affects the blood vessels. An HAE attack can result in rapid swelling of the hands, feet, limbs, face, intestinal tract, larynx (voicebox), or trachea (windpipe).

Attacks of swelling can become more severe in late childhood and adolescence.

There is usually a family history of the condition. But relatives may be unaware of previous cases, which may have been reported as an unexpected, sudden, and premature death of a parent, aunt, uncle, or grandparent.

Dental procedures, sickness (including colds and the flu), and surgery may trigger HAE attacks.

Symptoms

Symptoms include:

- Airway blockage -- involves throat swelling and sudden hoarseness
- Repeat episodes of abdominal cramping without obvious cause
- Swelling in the hands, arms, legs, lips, eyes, tongue, throat, or genitals
- Swelling of the intestines -- can be severe and lead to abdominal cramping, vomiting, dehydration, diarrhea, pain, and occasionally shock
- A non-itchy, red rash

Exams and Tests

Blood tests:

- C1 inhibitor function

- C1 inhibitor level
- Complement component 4 (may only be abnormal during an episode)

Treatment

Antihistamines, corticosteroids and other treatments used for idiopathic angioedema and hives (urticaria) do not work well for HAE. Epinephrine could be used in life-threatening reactions, although it is unlikely to stop the swelling. There are a number of newer FDA-approved treatments for HAE.

Some are given through a vein (IV) and can be used at home. Others are given as an injection under the skin by the person. Another is given by mouth. Some of these medicines are used regularly to prevent attacks and others are used emergently when an attack occurs to prevent it from getting worse. The choice of which agent may be based on the age of the person, personal preference, and where the symptoms occur.

Before these newer medicines became available, androgen medicines such as danazol, were used to reduce the frequency and severity of attacks. These medicines help the body make more C1 inhibitor. However, many women have serious side effects from these medicines. They can also not be used in children.

Once an attack occurs, treatment includes fast administration of specific medicines that stop the attack from worsening and supportive care such as pain relief and fluids given through a vein by an intravenous (IV) line.

Helicobacter pylori, a type of bacteria found in the stomach, can trigger abdominal attacks. Antibiotics to treat the bacteria help decrease abdominal attacks.

Support Groups

More information and support for people with HAE condition and their families can be found at:

- National Organization for Rare Disorders -- rarediseases.org/rare-diseases/hereditary-angioedema [https://rarediseases.org/rare-diseases/hereditary-angioedema]
- US Hereditary Angioedema Association -- www.haea.org [https://www.haea.org]

Outlook (Prognosis)

HAE can be life threatening and, despite new treatments, airway swelling can still lead to death. How well a person does depends on the specific symptoms.

Possible Complications

Swelling of the airways may be deadly.

When to Contact a Medical Professional

Contact or visit your health care provider if you are considering having children and have a family history of this condition. Also call if you have symptoms of HAE.

Swelling of the airway is a life-threatening emergency. If you are having difficulty breathing due to swelling, seek immediate medical attention.

Prevention

Genetic counseling may be helpful for prospective parents with a family history of HAE.

Alternative Names

Quincke disease; HAE - Hereditary angioedema; Kallikrein inhibitor - HAE; Bradykinin receptor antagonist - HAE; C1-inhibitors - HAE; Hives - HAE

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Review Date 3/31/2024

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