Hereditary Angioedema

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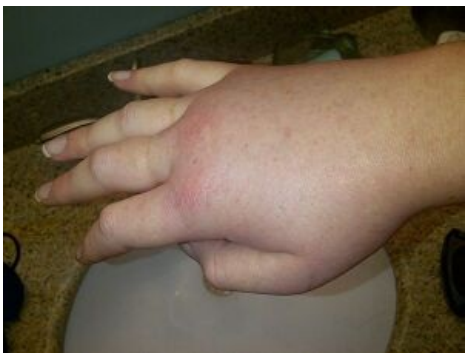
Hereditary angioedema (C1 inhibitor deficiency, OMIM: 106100 (http://omim.org/entry/106100)) is a genetic disease of the immune system with autosomal dominant inheritance. **This is not a classic immunodeficiency**, in which the affected individual would be exposed to a higher risk of infectious disease, but it is a hereditary disease of the immune system, where a mutation in the gene for one of the components of the immune system **damages its carrier**.



Swollen hand during a hereditary angioedema attack

Pathogenesis

The mutation affects **the complement component C1 inhibitor gene** (C1-INH, localization 11q11-q13.1). The product of the mutated gene is either not formed at all or is non-functional. This means that every stimulus (albeit minimal and harmless such as a minor injury) leads to the **activation** of the entire complement cascade, the clinical manifestations are responsible for increased **bradykinin** production, which is normally regulated by inhibition of prekallikrein to kallikrein conversion by C1-INH.

Clinical picture

Externally, the disease manifests itself in **swelling of the subcutaneous tissue and mucous membranes**, which are pale, itchy, without a local increase in temperature. Swelling of the **mucous membranes of the respiratory tract can be dangerous,** gastrointestinal mucosa can cause **indigestion**, in **severe form pain and partial intestinal obstruction** and mimic a sudden abdominal event, urinary tract mucosa is at risk of **urinary retention**.

The disease has a later onset, sometimes in adulthood.

Diagnosis

The diagnosis is based on the demonstration of l**ow levels of the C4 component of complement** (due to consumption) and **very low** (in the non-functional form of C1-INH also normal or high) **levels of C1-INH.**

Differential diagnosis

It is necessary to differentiate the acquired forms differentially - paraneoplastic (consumption of C1-INH in tumor tissue), parainfectious (rarely in Hellicobacter pylori infection) and drug (in ACEI therapy up to 0.3−1 %)

Therapy

The B2R receptor antagonist bradykinin is used in acute treatment.- icatibant s.c. and i.v. **C1-INH substitution**. **Antifibrinolytics** (tranexamic acid) and **danazol** (ethinyltestosterone derivative) are used for long-term prophylaxis.

Sources

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