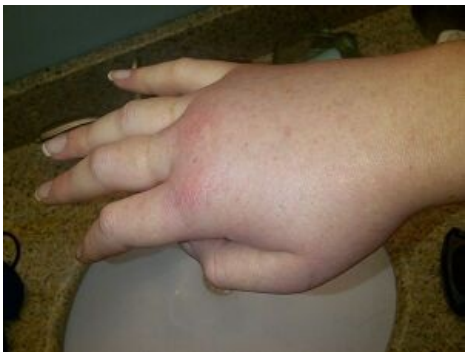
Hereditary angioedema

Hereditary angioedema (C1 inhibitor deficiency, OMIM: 106100) is a genetically determined disease of the immune system with autosomal dominant inheritance. **This is not a classic immunodeficiency** , where 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 .



A swollen hand in a patient with hereditary angioedema

Pathogenesis

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

Clinical picture

Externally, the disease is manifested by **swelling of the subcutaneous tissue** and mucous membranes , which are pale, non-itchy, without a local increase in temperature. **Swelling of the mucous membranes of the respiratory tract** can be **dangerous ,** damage to the mucous membranes of the digestive tract can manifest itself in **digestive difficulties** , in a more severe form, **pain and partial intestinal obstruction** and mimic a sudden abdominal attack, in the case of damage to the mucous membranes of the urinary tract, there is a risk of **urine retention .**

The disease has a later onset, sometimes in adulthood.

Diagnosis

The diagnosis is based on the evidence of **a low level of the C4** component of the complement (due to consumption) and **a very low** (in the case of a non-functional form of C1-INH and normal or high) **levels of C1-INH** .

Differential diagnosis

In terms of differential diagnosis, it is necessary to distinguish the acquired forms - paraneoplastic (consumption of C1-INH in the tumor tissue), parainfectious (rarely in *Hellicobacter pylori* infection) and drug-induced (in ACEI therapy in up to 0.3-1%).

Treatment

In acute treatment , B2R receptor antagonist for bradykinin − icatibant sc and iv **substitution of C1-INH** are used . Antifibrinolytics (tranexamic acid) and **danazol** (ethinyltestosterone derivative) are given as long-term prophylaxis

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- ŠIPEK, Antonín. *Genetically determined disorders of the immune system* [online]. Last revision 9/6/2006, [cit. 4/1/2010]. < http://www.genetika-biologie.cz/primarni-immunodeficiency >.

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