6. HEREDITARY ANGIOEDEMA WITH C1INHIBITOR DEFICIENCY

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Introduction. Hereditary angioedema (HAE) is a rare autosomal dominant disease with deficiency or/and dysfunction of C1 inhibitor, caused by mutations in the C1-INH gene. HAE is characterized by potentially life-threatening episodes of subcutaneous and/or submucosal edema without urticaria, prevalence 1:50,000 individuals. The primary mediator of swelling in HAE is bradykinin. The symptoms vary in severity, location, and duration, but the organs involved include the upper airways, skin, and gastrointestinal tract.

Case statement. A 19 y.o. man was admitted in the ICU with severe dyspnoea, bronchospasm, oedema of face and extremities, acute respiratory failure. Patient was diagnosed in 2018 with hereditary angioedema with C1Inh deficiency. Symptoms of severe angioedema (upper and lower limbs lasting for 2-3 days, abdominal attacks, nausea and vomiting, facial edema associated with difficult swallowing) started at age 2 years. The patient has frequent episodes of angioedema every month (predominantly limbs, face) caused by various triggers: stress, low/high temperatures, trauma and other factors. Sometimes marginal erythema may be present as a prodromal symptom. Investigations: C1 inhibitor 0,0411 g/l (N 0,21-0,4), C1 inhibitor function 0,0411% (N 70-130%), C3 0,86 g/l (N 0.9-1.8), C4 0.08 (N 0.1-0.4), d-Dimers 8,24 (0-0.55 mg FEU/l), qualitative troponin positive, CK-MB 96 (N 0 - 25 U/L), CRP 26 (0-5 mg/l). EKG, Echocardiography were normal. Treatment of choice: A plasma-derived C1 Esterase Inhibitor, fresh frozen plasma and tracheal intubation in severe cases (laryngeal angioedema). Patient was discharged on the 10th day with improvement of general condition with recommendation on management and prevention of further HAE attacks.

Discussions. Because the disease is very rare, it is not uncommon for patients to remain undiagnosed for many years. Diagnostic delays impact the accuracy of management. Treatment is different from histamine-associated angioedema, antihistamines, corticosteroids, and epinephrine have no effect. Acute treatment of HAE can include IV infusions of C1-INH, receptor antagonists, and kallikrein inhibitors. Short- and long-term prophylaxis can also be administered to patients with HAE.

Conclusion. In patients with early onset of repetitive angioedema episodes, not responding to antihistamines, corticosteroids therapy the diagnosis of HAE should be considered.