C1 Esterase Inhibitor Deficiency

PLAY PICMONIC

Pathophysiology

Autosomal Dominant
Domino
This condition is inherited in an autosomal dominant pattern. This means that the inheritance of one abnormal allele is enough for the child to manifest the disorder.

Disinhibition of Kallikrein
Disinhibited Crazy Calico Cat On Crane
C1 esterase inhibitor (C1INH)inhibits kallikrein, which is responsible for the degradation of low molecular weight kininogen into bradykinin. When C1 esterase inhibitor is deficient, kallikrein is disinhibited.

Increased Bradykinin
Up-arrow Brady-crying
Under normal circumstances, C1 esterase inhibitor inhibits kallikrein, which is responsible for the degradation of low molecular weight kininogen into bradykinin. When C1 esterase inhibitor is deficient, kallikrein is upregulated, and conversion of low molecular weight kininogen into bradykinin increases. This results in increased levels of bradykinin.

Decreased C4
Down-arrow Cat-(4)-fork
Disinhibition of C1 esterase promotes cleavage and depletion of C4. Decreased C4 concentration in the serum is common in these patients and can aid in making the diagnosis.

Clinical Features

Recurrent Angioedema
Angel-edamame
C1 esterase deficiency causes hereditary angioedema, which is an autosomal dominant condition characterized by recurrent episodes of severe swelling (angioedema). The edema usually involves deeper dermis and subcutaneous tissue. The swelling may occur in the abdomen, arms, legs, face, tongue, and larynx. Obstruction of the airways can result in a quick death.

Upper Airway Obstruction
Obstructed-Airway
Increased bradykinin levels lead to soft tissue edema. The most feared complication of C1 esterase inhibitor deficiency is laryngeal edema as it can cause upper airway obstruction and death.

Considerations

ACE Inhibitors Contraindicated

ACE with inhibiting-chains Caution-Tape

ACE inhibitors increase bradykinin levels and are contraindicated in these patients as they have high bradykinin levels at the baseline, and a further increase in bradykinin levels can lead to cough and paroxysms of angioedema.

Avoid Triggers

Avoid-sign with Trigger

Symptoms are commonly triggered by trauma, psychological stress, food (e.g., shellfish, nuts), infections, etc. Individual triggers should be identified and avoided.