Opis choroby *

Definicja

A rare hereditary angioedema characterized by potentially life-threatening episodes of subcutaneous and/or submucosal edema without urticaria and with normal levels and function of C1 esterase inhibitor. Patients present with prolonged attacks which last for approximately two to five days and may include nonpitting edema of the skin, severe abdominal symptoms such as pain and swelling, and/or respiratory distress due to upper respiratory airways involvement. Affected locations and frequency of attacks differ slightly between subtypes. Estrogen-containing oral contraceptives and pregnancy are precipitating factors, especially in patients with a factor XII mutation.

Dane

Klasyfikacja
Choroba

Synonimy
HAE with normal C1 inhibitor
HAE z prawidłowym poziomem C1Inh
HAE z prawidłowym poziomem inhibitora C1
Dziedziczny obrzęk naczynioruchowy z prawidłowym poziomem inhibitora C1
Dziedziczny obrzęk angioneurotyczny z prawidłowym poziomem C1Inh
Dziedziczny obrzęk angioneurotyczny z prawidłowym poziomem inhibitora C1
HAE with normal C1Inh
Hereditary angioedema with normal C1 inhibitor
Hereditary angioneurotic edema with normal C1 inhibitor
Hereditary angioneurotic edema with normal C1Inh

Kod ORPHA
528647

Kod OMIM
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Kod ICD10
T78.3

Kod ICD11
4A00.14

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*Źródło

orphanet