

Opis choroby *

Definicja

A rare hereditary angioedema characterized by potentially life-threatening episodes of subcutaneous and/or submucosal edema without urticaria, associated with C1 esterase inhibitor (C1-INH) deficiency. Hereditary angioedema (HAE) type 1 is caused by quantitative, HAE type 2 by qualitative defects of C1-INH. The two subtypes are clinically indistinguishable. Patients may present at any age (but most commonly in childhood) with recurrent attacks of nonpitting edema of the skin, severe abdominal symptoms such as pain and swelling, and/or respiratory distress due to upper respiratory airways involvement. Genital, bladder, muscle, or joint swelling may occur in some cases.

Dane

Klasyfikacja

Choroba

Synonimy

HAE with C1 inhibitor deficiency
HAE z niedoborem C1Inh
HAE z niedoborem inhibitora C1
Dziedziczny obrzęk angioneurotyczny z niedoborem C1Inh
Dziedziczny obrzęk angioneurotyczny z niedoborem inhibitora C1
HAE with C1Inh deficiency
Hereditary angioneurotic edema with C1 inhibitor deficiency
Hereditary angioneurotic edema with C1Inh deficiency

Kod ORPHA

528623

Kod OMIM

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Kod ICD10

D84.1

Kod ICD11

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*Źródło

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