Dziedziczny obrzęk naczynioruchowy z niedoborem C1Inh
Kod Orpha: 528623 Kod OMIM:

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

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.