

# Dziedziczny obrzęk naczyń ruchowy 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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## **Rozszerzony opis choroby**

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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