

# Niedobór odporności z powodu wadliwej ekspresji HLA klasy 1

## Kod Orpha: 34592 Kod OMIM: 241600

### Opis choroby \*

#### Definicja

A rare autosomal recessive primary immunodeficiency characterized by severe reduction in the cell surface expression of HLA class I molecules, typically resulting in childhood-onset of chronic bacterial infections of the respiratory tract evolving to widespread bronchiectasis and respiratory insufficiency. Sterile necrotizing granulomatous skin lesions mainly involving the extremities and the mid-face may be observed in some patients. Severe viral infections do not occur as part of the condition. Atypical variants without respiratory or cutaneous manifestations, as well as asymptomatic individuals have been reported.

#### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Bare lymphocyte syndrome type 1  
Bare lymphocyte syndrome type 1  
MHC class I deficiency

#### Kod ORPHA

34592

#### Kod OMIM

241600

#### Kod ICD10

D81.6

#### Kod ICD11

4A01.11

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#### [\\*Źródło](#)

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### Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

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