

## Opis choroby \*

### Definicja

A rare genetic immune disease characterized by early onset of recurrent bacterial, viral, and fungal infections, chronic inflammatory bowel disease, gastritis, and inflammatory polyarthritis. Patients present with diarrhea, vomiting, hepatosplenomegaly, mouth ulcers, perianal abscesses, chronic lung disease with bronchiectasis, and failure to thrive. Occurrence of a skin rash associated with lymphocytic vasculitis has also been reported. Immunologic abnormalities include variable T-cell lymphopenia, decreased natural killer cells, and decreased B-cells with variable hypogammaglobulinemia.

### Dane

### Klasyfikacja

Choroba

#### Kod ORPHA

529977

#### Kod OMIM

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#### Kod ICD10

D89.8

#### Kod ICD11

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### \*Źródło

orphonet