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
-

Kod ICD10
D89.8

Kod ICD11
-

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*Źródło

orphanet