

## Opis choroby \*

### Definicja

A rare, genetic, non-severe combined immunodeficiency disease characterized by immunodeficiency (manifested by recurrent and/or severe bacterial and viral infections), destructive noninfectious granulomas involving skin, mucosa and internal organs, and various autoimmune manifestations (including cytopenias, vitiligo, psoriasis, myasthenia gravis, enteropathy). Immunophenotypically, T-cell and B-cell lymphopenia, hypogammaglobulinemia, abnormal specific antibody production and impaired T-cell function are observed.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

CID due to RAG 1/2 deficiency

CID z powodu niedoboru RAG 1/2

Złożony Niedobór odporności z powodu niedoboru RAG 1/2

Combined immunodeficiency due to RAG 1/2 deficiency

#### Kod ORPHA

157949

#### Kod OMIM

233650

#### Kod ICD10

D81.1

#### Kod ICD11

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#### \*Źródło

orphanet