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