Opis choroby *

Definicja

Severe combined immunodeficiency due to complete RAG1/2 deficiency is a rare, genetic T-B- severe combined immunodeficiency disorder due to null mutations in recombination activating gene (RAG) 1 and/or RAG2 resulting in less than 1% of wild type V(D)J recombination activity. Patients present with neonatal onset of life-threatening, severe, recurrent infections by opportunistic fungal, viral and bacterial micro-organisms, as well as skin rashes, chronic diarrhea, failure to thrive and fever. Immunologic observations include profound T- and B-cell lymphopenia, normal NK counts and low or absent serum immunoglobulins; some patients may have eosinophilia.

Dane

Klasyfikacja
Choroba

Synonimy
SCID due to complete RAG1/2 deficiency
SCID z powodu całkowitego niedoboru RAG1/2

Kod ORPHA
331206

Kod OMIM
601457

Kod ICD10
D81.1

Kod ICD11
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*Źródło

orphanet