Opis choroby *

Definicja

A rare severe combined immunodeficiency characterized by T-cell lymphopenia and absent T-cell proliferative responses, and normal B-cell and natural killer cell counts. Patients present in the first months of life with severe recurrent infections, failure to thrive, hematologic autoimmune disorders, and/or lymphoproliferation with splenomegaly.

Dane

Klasyfikacja
Choroba

Synonimy
SCID due to LAT deficiency
SCID due to LAT deficiency

Kod ORPHA
504523

Kod OMIM
617514

Kod ICD10
D81.2

Kod ICD11
4A01.10

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*Źródło

orphanet