

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

*Źródło

orphanet