

Opis choroby *

Definicja

A rare primary immunodeficiency with autosomal or X-linked recessive inheritance, characterized by atrophy of the thymus in the absence of other congenital abnormalities, with profound T-cell deficiency, while serum immunoglobulin levels are normal or increased. Patients present with chronic or recurrent infections in infancy including candidiasis, skin, pulmonary and urinary tract infections, chronic diarrhea, and failure to thrive.

Dane

Klasyfikacja

Choroba

Synonimy

Nezelof syndrome

Zespół Nezelofa

Niedobór odporności zależnej od limfocytów T z aplazją grasicy

Kod ORPHA

83471

Kod OMIM

242700

Kod ICD10

D81.4

Kod ICD11

4A01.30

[*Źródło](#)

orphanet