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