Opis choroby *

Definicja

A rare autosomal recessive primary immunodeficiency characterized by partial T lymphopenia (in particular cytotoxic CD8+ cells) and decreased expression of the T cell receptor (TCR)/CD3 complex with impaired proliferative response to TCR-dependent stimuli, while the mature memory T cell pool is comparatively well preserved, and B cells, natural killer cells, and immunoglobulins are typically normal. The clinical phenotype is highly heterogeneous, ranging from asymptomatic to infancy-onset of severe recurrent infections, as well as occurrence of autoimmune disease or enteropathy.

Dane

Klasyfikacja
Choroba

Kod ORPHA
169082

Kod OMIM
615607

Kod ICD10
D81.2

Kod ICD11
4A01.1Y

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*Źródło

orphanet