

Opis choroby *

Definicja

A rare primary immunodeficiency disorder due to impaired capacity of activated T- and B-cells to proliferate in response to antigen receptor-mediated activation characterized by early-onset, severe, persistent and/or recurrent viral infections due to Epstein-Barr virus (EBV) and Varicella Zoster virus (VZV, including generalized varicella), as well as recurrent sino-pulmonary bacterial infections due to encapsulated pathogens.

Dane

Klasyfikacja

Choroba

Synonimy

SCID due to CTPS1 deficiency

SCID z powodu niedoboru CTPS1

Kod ORPHA

420573

Kod OMIM

615897

Kod ICD10

D81.2

Kod ICD11

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*Źródło

orphanet