

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 -		

*Źródło

orphanet