Opis choroby *

Definicja

A rare genetic immune disease characterized by infantile or childhood onset of combined immunodeficiency with recurrent viral, bacterial, and fungal infections, severe autoimmunity mainly manifesting as antibody-mediated destruction of red blood cells, platelets, and neutrophils, and mild to moderate developmental delay. Laboratory findings include decreased circulating T-, B-, and natural killer cells, and hypergammaglobulinemia.

Dane

Klasyfikacja
Choroba

Synonimy
Evans syndrome associated with primary immunodeficiency
Autoimmunologiczna niedokrwistość hemolityczna i małopłytkowość autoimmunologiczna z pierwotnym niedoborem odporności
Choroba TRIANGLE
Niedobór odporności zależny od TPPII, autoagresja, opóźnienie rozwoju nerwowego z upośledzeniem glikolizy i ekspansą lyzosomów
TPPII deficiency
TPPII-related immunodeficiency, autoimmunity, and neurodevelopmental delay with impaired glycolysis and lysosomal expansion disease
TRIANGLE disease
Tripeptidyl-peptidase II deficiency

Kod ORPHA
444463

Kod OMIM
619220

Kod ICD10
D89.8

Kod ICD11
-

---

*Źródło

orphanet