## Opis choroby *

Definicja

A rare immune dysregulation disease with immunodeficiency characterized by infantile or childhood onset of a variable phenotype including recurrent/persistent bacterial, fungal, and viral infections with involvement of the skin, lower respiratory tract, and gastrointestinal tract, eczema, allergies, and inflammatory bowel disease, among others. EBV-related smooth muscle tumors have also been reported. Immunophenotyping shows decreased Treg counts, as well as a deficient CD3/CD28 co-stimulation response in CD4+ and CD8+ T-cells.

Dane

**Klasyfikacja**
Choroba

Synonimy
Combined immunodeficiency due to RLTPR deficiency
Ciężki złożony niedobór odporności z powodu niedoboru RLTPR

**Kod ORPHA**
542301

**Kod OMIM**
618131

**Kod ICD10**
D82.3

**Kod ICD11**
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*Źródło

orphanet