

## 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