

## Opis choroby \*

### Definicja

A rare, genetic, primary immunodeficiency characterized by early onset of recurrent respiratory infections and variable combination of autoimmune disorders, including hemolytic anemia, thrombocytopenic purpura, lymphoproliferative disease, inflammatory bowel disease, colitis, diabetes, arthritis, and dermatitis. Failure to thrive, hepatosplenomegaly and endocrine abnormalities have also been associated. Variable immunologic findings include deficiency of CD4+ T regulatory cells, decreased B-cells, and hypogammaglobulinemia.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

CID due to LRBA deficiency

CID spowodowany niedoborem LRBA

#### Kod ORPHA

445018

#### Kod OMIM

614700

#### Kod ICD10

D81.8

#### Kod ICD11

4A01.21

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#### \*Źródło

orphanet