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