Opis choroby *

Definicja

A rare autosomal recessive primary immunodeficiency characterized by absence of HLA class II molecules on the surface of immune cells, leading to severely impaired cellular and humoral immune response to foreign antigens, severe CD4+ T-cell lymphopenia, and hypogammaglobulinemia. The disease clinically manifests with early onset of severe and recurrent infections mainly of the respiratory and gastrointestinal tract, protracted diarrhea with failure to thrive, and autoimmune disease, and is frequently fatal in childhood.

Dane

Klasyfikacja
Choroba

Synonimy
Bare lymphocyte syndrome type 2
Ciężki złożony Niedobór odporności HLA klasy 2
Zespół nagich limfocytów typu 2
MHC class II deficiency

Kod ORPHA
572

Kod OMIM
209920

Kod ICD10
D81.7

Kod ICD11
4A01.12

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*Źródło

orphanet