Opis choroby *

Definicja

A rare autosomal recessive primary immunodeficiency characterized by infancy onset of severe inflammatory bowel disease with life-threatening diarrhea and failure to thrive, oral aphthous ulcers, and recurrent severe upper and lower respiratory tract infections with finger clubbing. Laboratory examination reveals increased IgE and decreased IgG levels, as well as reduced numbers of circulating CD19+ B-cells including IgM+ naive and class-switched IgG memory B-cells, with a concomitant increase in transitional B-cells, while T-cell numbers and function are normal.

Dane

Klasyfikacja
Choroba

Synonimy
IL21-related infantile IBD
Niemowlęce IBD zależna od IL21

Kod ORPHA
477661

Kod OMIM
615767

Kod ICD10
D84.8

Kod ICD11
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*Źródło

orphanet