Opis choroby *

Definicja

A rare, genetic, non-severe combined immunodeficiency disorder characterized by variable B- and T-cell defects (including defective B-cell differentiation and impaired T-cell proliferation to mitogens and bacterial antigens) and natural killer cell dysfunction (ranging from impaired cytotoxicity to lymphopenia) due to IL21R deficiency, manifesting with recurrent respiratory and/or gastrointestinal tract infections and, in some cases, with severe, chronic, progressive cholangitis and liver cirrhosis associated with cryptosporidial infection.

Dane

Klasyfikacja
Choroba

Kod ORPHA
357329

Kod OMIM
615207

Kod ICD10
D81.8

Kod ICD11
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*Źródło

orphanet