Opis choroby *

Definicja

A rare genetic combined T and B cell immunodeficiency characterized by life-threatening infections due to disrupted transferrin receptor 1 endocytosis, resulting in defective cellular iron transport and impaired T and B cell function. Patients present with early-onset chronic diarrhea, severe recurrent infections, and failure to thrive. Laboratory studies reveal hypo- or agammaglobulinemia, normal lymphocyte counts but decreased numbers of memory B cells, intermittent neutropenia and thrombocytopenia, and mild anemia (resistant to iron supplementation) with low mean corpuscular volume.

Dane

Klasyfikacja
Choroba

Synonimy
CID due to TFRC deficiency
TFRC-related combined immunodeficiency
CID due to TFRC deficiency
TFRC-related combined immunodeficiency

Kod ORPHA
476113

Kod OMIM
616740

Kod ICD10
D81.8

Kod ICD11
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*Źródło

orphanet