

## 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