# Złożony niedobór odporności z powodu niedoboru GINS1
# Kod Orpha: 505227 Kod OMIM: 617827

## Opis choroby *

**Definicja**
A rare syndrome with combined immunodeficiency characterized by intrauterine and postnatal growth retardation, chronic neutropenia, and natural killer (NK) cell deficiency due a defect in DNA replication leading to blockade of immune cell differentiation in the bone marrow, particularly affecting NK cells. Other clinical features include recurrent viral and bacterial infections and eczema, as well as mild facial dysmorphism.
**Dane**

**Klasyfikacja**
Choroba

**Synonimy**
CID due to GINS1 deficiency
Combined immunodeficiency with intrauterine growth retardation-NK cell deficiency-neutropenia
Combined immunodeficiency with intrauterine growth retardation-natural killer cell deficiency-neutropenia
CID due to GINS1 deficiency
Combined immunodeficiency with intrauterine growth retardation-NK cell deficiency-neutropenia
Combined immunodeficiency with intrauterine growth retardation-natural killer cell deficiency-neutropenia

**Kod ORPHA**
505227

**Kod OMIM**
617827

**Kod ICD10**
D81.8

**Kod ICD11**
4A01.1Y

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*Źródło

orphanet

## Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.