

## 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