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