

Opis choroby *

Definicja

A rare primary immunodeficiency characterized by regular oscillations in blood neutrophil counts from normal or subnormal levels to severe neutropenia, usually with a cycle length of about 21 days. Symptoms during the neutropenic phase include fever, mouth ulcers, but also pneumonia, and peritonitis, among others. Mode of inheritance is autosomal dominant.

Dane

Klasyfikacja

Choroba

Kod ORPHA

2686

Kod OMIM

162800

Kod ICD10

D70

Kod ICD11

4B00.00

*Źródło

orphanet