

# **Autosomalna recesywna ciężka wrodzona neutropenia z powodu niedoboru CSF3R**

## **Kod Orpha: 420702 Kod OMIM: 617014**

### **Opis choroby \***

#### **Definicja**

Autosomal recessive severe congenital neutropenia due to CSF3R deficiency is a rare, genetic, primary immunodeficiency disorder characterized by predisposition to recurrent, life-threatening bacterial infections associated with decreased peripheral neutrophil granulocytes (absolute neutrophil count less than 500 cells/microliter), resulting from recessively inherited loss-of-function mutations in the *CSF3R* gene. Full maturation of all three lineages in the bone marrow and refractoriness to *in vivo* rhG-CSF treatment are associated.

#### **Dane**

#### **Klasyfikacja**

Choroba

**Kod ORPHA**  
420702

**Kod OMIM**  
617014

**Kod ICD10**  
D70

#### **Kod ICD11**

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[\\*Źródło](#)

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### **Rozszerzony opis choroby**

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.

Dostępna na stronie [www.orphanet.pl](http://www.orphanet.pl)