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
-

*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.