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

orphanet