Autosomal recessive severe congenital neutropenia due to CSF3R deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive severe congenital neutropenia due to CSF3R deficiency. ORPHA:420702

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.