sciensano **Belgian Genetic Tests database**

DISEASE:
Autosomal recessive severe congenital neutropenia due to CSF3R deficiency

NAME:	Autosomal recessive severe congenital neutropenia due to CSF3R deficiency
DESCRIPTION:	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.
ORPHACODE:	420702
XREF(S):	Orphanet OMIM ICD-10
ANALYTE(S):	CSF3R
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