DISEASE:
Autosomal dominant severe congenital neutropenia

NAME:	Autosomal dominant severe congenital neutropenia
DESCRIPTION:	A rare primary immunodeficiency disorder characterized by autosomal dominant inheritance, absolute neutrophil counts below 0.5x10E9/L in the peripheral blood (on three separate occasions over a six month period), granulopoiesis maturation arrest at the promyelocyte/myelocyte stage and early-onset, severe, recurrent bacterial infections.
ORPHACODE:	486
XREF(S):	Orphanet OMIM OMIM ICD-10 OMIM OMIM OMIM

ANALYTE(S):	TCIRG1 ELANE GFI1 CLPB SRP19 SRP54
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Myeloid/lymphoid neoplasms with germline predisposition (gene panel)
- Primary immune deficiencies (gene panel)
- « Inherited bone marrow failures syndromes » with or without organ dysfunction

Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- ClpB family mitochondrial disaggregase
- elastase, neutrophil expressed
- growth factor independent 1 transcriptional repressor
- signal recognition particle 19
- signal recognition particle 54
- T cell immune regulator 1, ATPase H+ transporting V0 subunit a3

Related Gene Panels

- Hematologic Familiar Forms - ULG
- Primary immune deficiencies (444 genes) - KUL
