

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 $0.5 \times 10^9/L$ 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):	<u>TCIRG1</u> <u>ELANE</u> <u>GFI1</u> <u>CLPB</u> <u>SRP19</u> <u>SRP54</u>
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