DISEASE:
Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency

NAME:	Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency
DESCRIPTION:	Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency is a rare, genetic, primary immunodeficiency disorder characterized by early-onset, recurrent, severe bacterial infections, granulopoiesis maturation arrest at the promyelocyte/myelocyte stage and markedly reduced absolute neutrophil counts, resulting from recessively inherited mutations in the JAGN1 gene. Mild facial dysmorphism (i.e. triangular face), short stature, failure to thrive, hypothyroidism, developmental delay, pancreatic insufficiency and coarctation of aorta, as well as bone and urogenital abnormalities, may also be associated.
ORPHACODE:	423384
XREF(S):	Orphanet ICD-10 OMIM
ANALYTE(S):	JAGN1
CREATED:	13 May 2019 - 01:02
CHANGED:	22 Jun 2023 - 16:14

RELATED CONTENT

Related Genetic Tests

- Primary immune deficiencies (gene panel)

Related Laboratories

- Centrum Menselijke Erfelijkheid - KUL

Related Analytes

- jagunal homolog 1

Related Gene Panels

- Primary immune deficiencies (444 genes) - KUL