

Opis choroby *

Definicja

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.

Dane

Klasyfikacja

Choroba

Kod ORPHA

423384

Kod OMIM

616022

Kod ICD10

D70

Kod ICD11

4B00.00

*Źródło

orphanet