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

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*Źródło

orphanet