Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Autosomal recessive severe congenital neutropenia due to JAGN1 deficiency. ORPHA:423384

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 coractation of aorta, as well as bone and urogenital abnormalities, may also be associated.