Opis choroby *

Definicja

Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency is a rare, genetic, primary immunodeficiency disorder characterized by increased susceptibility to recurrent, life-threatening bacterial infections, in association with typically severe neutropenia in peripheral blood and bone marrow and a prominent ectatic superficial vein pattern, resulting from recessively inherited mutations in the G6PC3 gene. Cardiac malformations (e.g. atrial septal defects, patent ductus arteriosus,valvular defects), urogenital anomalies (incl. cryptorchidism), growth and developmental delay, facial dysmorphism (e.g. frontal bossing, upturned nose, malar hypoplasia), and intermittent thrombocytopenia are frequently associated.

Dane

Klasyfikacja
Choroba

Synonimy
SCN4
Ciężka wrodzona neutropenia - nadciśnienie płucne - powierzchowna angiektazja żylna
Ciężka wrodzona neutropenia 4
SCN4
Severe congenital neutropenia type 4
Severe congenital neutropenia-pulmonary hypertension-superficial venous angiectasis syndrome

Kod ORPHA
331176

Kod OMIM
612541

Kod ICD10
D70

Kod ICD11
4B00.00

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*Źródło

orphanet