# DISEASE:
# Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency

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| **NAME:** | Autosomal recessive severe congenital neutropenia due to G6PC3 deficiency |
| **DESCRIPTION:** | 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. |
| **ORPHACODE:** | 331176 |
| **SYNONYMS:** | SCN4<br>Severe congenital neutropenia type 4<br>Severe congenital neutropenia-pulmonary hypertension-superficial venous angiectasis syndrome |
| **XREF(S):** | Orphanet<br>ICD-10<br>OMIM |
| **ANALYTE(S):** | G6PC3 |
| **CREATED:** | 13 May 2019 - 01:02 |

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| **CHANGED:** | 22 Jun 2023 - 16:14 |

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# RELATED CONTENT

## Related Genetic Tests

- Primary immune deficiencies (gene panel)
- « Inherited bone marrow failures syndromes » with or without organ dysfunction

## Related Laboratories

- Centre de Génétique Humaine - CHU Sart-Tilman
- Centrum Menselijke Erfelijkheid - KUL

## Related Analytes

- glucose-6-phosphatase catalytic subunit 3

## Related Gene Panels

- Hematologic Familiar Forms - ULG
- Primary immune deficiencies (444 genes) - KUL