Wrodzona ciężka neutropenia sprzężona z chromosomem X
Kod Orpha: 86788 Kod OMIM: 300299

Opis choroby *

Definicja
X-linked severe congenital neutropenia is an immunodeficiency syndrome characterized by recurrent major bacterial infections, severe congenital neutropenia, and monocytopenia. It has been described in five males spanning three generations of one family. It is transmitted as an X-linked recessive trait and is caused by mutations in the WAS gene, encoding the WASP protein.
Dane

Klasyfikacja
Choroba

Kod ORPHA
86788

Kod OMIM
300299

Kod ICD10
D70

Kod ICD11
4B00.00

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*Źródło

orphanet

Rozszerzony opis choroby

Brak opisu rozszerzonego dla tej choroby. Opracowanie w toku.