

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

*Źródło

orphanet