

## 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