

## Opis choroby \*

### Definicja

A rare disorder characterized by the association of severe combined immunodeficiency (affecting mainly the humoral immune response) with progressive cerebellar ataxia. It is characterized by neurological signs, telangiectasia, increased susceptibility to infections and a higher risk of cancer.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

Louis-Bar syndrome

„Happy puppet” syndrome

zespół Louis-Barr

niezborność z poszerzeniem naczyń

powierzchnowych

AT

#### Kod ORPHA

100

#### Kod OMIM

208900, 208910, 607585

#### Kod ICD10

G11.3

#### Kod ICD11

4A01.31

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#### \*Źródło

orphanet