

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

niezbornosć z poszerzeniem naczyń

powierzchnowych

AT

Kod ORPHA

100

Kod OMIM

208900, 208910, 607585

Kod ICD10

G11.3

Kod ICD11

4A01.31

*Źródło

orphanet