

Opis choroby *

Definicja

A rare immune disease characterized by progressive immunodeficiency leading to recurrent and opportunistic infections, autoimmunity and malignancy as well as neurologic manifestations.

Dane

Klasyfikacja

Choroba

Synonimy

PNP deficiency

Niedobór PNP

Niedobór PNPazy

PNPase deficiency

Kod ORPHA

760

Kod OMIM

613179

Kod ICD10

D81.5

Kod ICD11

4A01.1Y

*Źródło

orphanet