

## 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

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#### \*Źródło

orphanet