

## Opis choroby \*

### Definicja

A rare X-linked disorder of purine metabolism associated with hyperuricemia and hyperuricosuria, and comprised of two forms: an early-onset severe form characterized by gout, urolithiasis, and neurodevelopmental anomalies and a mild late-onset form with no neurologic involvement.

### Dane

#### Klasyfikacja

Choroba

#### Synonimy

PRPP synthetase superactivity

Nadczynność syntetazy PRPP

PRPS1 superactivity

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#### Kod ORPHA

3222

#### Kod OMIM

300661

#### Kod ICD10

E79.8

#### Kod ICD11

5C55.0Y

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#### \*Źródło

orphanet