

## Opis choroby \*

### Definicja

A severe form of phosphoribosylpyrophosphate (PRPP) synthetase superactivity, an X-linked disorder of purine metabolism, characterized by early onset hyperuricemia and hyperuricosuria, and clinically manifesting with urolithiasis, gout and neurodevelopmental anomalies consisting of variable combinations of sensorineural hearing loss, hypotonia, and ataxia.

### Dane

#### Klasyfikacja

Podtyp kliniczny

#### Synonimy

Severe PRPP synthetase superactivity  
Ciężka nadczynność PRPS1  
Ciężka nadczynność syntetazy PRPP  
Severe PRPS1 superactivity

#### Kod ORPHA

411543

#### Kod OMIM

300661

#### Kod ICD10

E79.8

#### Kod ICD11

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#### \*Źródło

orphanet