

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

PRPS1 superactivity

Kod ORPHA

3222

Kod OMIM

300661

Kod ICD10

E79.8

Kod ICD11

5C55.0Y

*Źródło

orphanet