

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	Synonimy
Choroba	PRPP synthetase superactivity Nadczynność syntetazy PRPP PRPS1 superactivity PRPS1 superactivity
Kod ORPHA	Kod OMIM
3222	300661
Kod ICD11	Kod ICD10
5C55.0Y	E79.8

*Źródło

orphanet