

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	Synonimy
Podtyp kliniczny	Severe PRPP synthetase superactivity Ciężka nadczynność PRPS1 Ciężka nadczynność syntetazy PRPP Severe PRPS1 superactivity

Kod ORPHA	Kod OMIM	Kod ICD10
411543	300661	E79.8

Kod ICD11

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*Źródło

orphanet