

Opis choroby *

Definicja

A rare disorder of proline metabolism characterized biochemically by markedly elevated levels of proline in plasma and urine due to deficiency of proline oxidase. The reported clinical phenotype ranges from asymptomatic to variable neurologic and psychiatric manifestations (including global developmental delay, seizures, autistic features, and hyperactivity).

Dane

Klasyfikacja

Choroba

Synonimy

Proline oxidase deficiency

Niedobór oksydazy prolinowej

Kod ORPHA

419

Kod OMIM

239500

Kod ICD10

E72.5

Kod ICD11

5C50.8

*Źródło

orphanet