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

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*Źródło

orphanet