## 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