Opis choroby *

Definicja

A rare inborn error of metabolism characterized by elevated levels of imino acids (proline, hydroxyproline) and glycine in urine due to defective reabsorption in the kidney. The condition is considered benign and not associated with any specific clinical phenotype. Mode of inheritance is autosomal recessive.

Dane

Klasyfikacja
Choroba

Kod ORPHA
42062

Kod OMIM
242600

Kod ICD10
E72.0

Kod ICD11
5C60.Y

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*Źródło

orphanet