

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

*Źródło

orphanet