Opis choroby *

Definicja

A rare inborn error of metabolism characterized by abnormal accumulation of plasma cystathionine and subsequent increased urinary excretion due to cystathionine gamma-lyase deficiency. The condition is considered benign without pathological relevance. Mode of inheritance is autosomal recessive.

Dane

Klasyfikacja
Choroba

Synonimy
Cystathionase deficiency
Niedoboóe gamma-cystationazy
Niedobór cystationazy
Niedobór liazy gamma - cystationu
Cystathionine gamma-lyase deficiency syndrome
Gamma-cystathionase deficiency

Kod ORPHA
212

Kod OMIM
219500

Kod ICD10
E72.1

Kod ICD11
5C50.B

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*Źródło

orphanet